cystinosis magazine

For friends and supporters of the Cystinosis Research Foundation

CYSTINOSIS COMMUNITY CHAMPIONS

SPRING 2016

HOPE THROUGH

COMMUNITY

ISSUE

Lars and Sam Jenkins.

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ON THE COVER:
Cystinosis Community Champion, musician Mindy Gledhill
Photo by: www.jefrastarrlinn.com
Inset: Sam and Lars Jenkins
See story on page 8.

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CYSTINOSIS RESEARCH FOUNDATION
RESEARCH • HOPE • CURE

The mission of the Cystinosis Research Foundation is to find better treatments and a cure for cystinosis by supporting bench, clinical and translational research. Since 2003, CRF has raised $33 million for cystinosis research in an effort to find a cure.

WELCOME

CRF CHAMPIONS

RESEARCH HIGHLIGHTS

ANNOUNCEMENTS

FAMILY STORIES

CYSTINOSIS MAGAZINE IS A PUBLICATION OF THE CYSTINOSIS RESEARCH FOUNDATION

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Dear Family & Friends

In this issue of Cystinosis Magazine we celebrate the champions in our lives – the people who have no family connection to the cystinosis community – yet they have embraced our cause and work tirelessly on behalf of this very small group of special people who desperately need our help. These champions, from every part of the world and from every walk of life, have made our quest for a cure their quest too.

On the cover is a beautiful picture of Mindy Gledhill, a singer and songwriter, who after hearing about Sam and Lars Jenkins from Salt Lake City, Utah, and their battle with cystinosis, performed a concert in their honor, donating the proceeds to cystinosis research. As a community, we are surrounded by wonderful people like Mindy. Some we know well, and others we have just met, but each of them has chosen to support our children by dedicating their time, energy and resources to help us find a cure for cystinosis. These champions, abundant with courage and conviction, lift us up and share our journey because they care and because they want to make a difference. The Cystinosis Research Foundation could not have achieved what it has without the support of these champions and all of you.

A Night of Incredible Surprises!

On April 9, a record-breaking crowd of more than 485 people joined us for the 2016 Natalie's Wish Celebration. Award-winning singer and songwriter Rachel Platten set the perfect tone for the evening when she performed her powerful anthem, Fight Song, during her opening set. Surrounded by cystinosis children who enthusiastically sang along, Rachel's lyrics could not have been more on point. It was truly a magical moment during a night filled with them.

This year Lauren Hartz from Pennsylvania spoke from her heart about her son Landon's life with cystinosis. Her heartbreaking and somehow still uplifting talk brought tears to the eyes of many guests as she shared the realities of cystinosis and the hopes that she and her husband, Jimmy, have for Landon as we journey to a cure.

The most touching aspect of the evening was knowing that 55 cystinosis families and adults with cystinosis from around the world had joined us for the event. Many of the families raise funds for CRF and cystinosis research in their own communities. It is always a highlight of our celebration when those families proudly parade on stage to present their checks. We are a small community but we are strong and united, and on this night we raised more than $3.3 million to bring our goal of a cure closer than ever to reality.

The Realities of Cystinosis

It seems like only yesterday that Natalie made her wish, "to have my disease go away forever." Much has happened since then to improve the lives of those with cystinosis and today, we have more hope than ever before. We do not have a cure but we are close.

We have lived with cystinosis since Natalie was an infant and now we have an adult child with the disease. Although Natalie is an adult and independent, she will always be my child. As her mother, all I want to do is shelter her from the realities of this disease. As our children grow into adulthood, there are more challenges, not less. Cystinosis does not pause along the way or reverse itself as our children grow older. The disease advances slowly, painfully and relentlessly. I am sure I am like many other parents of children with cystinosis – every day I wish I could take her place and give her even just one day free of pain and worry – a day when she does not have to think about or deal with cystinosis.

Even though Natalie does not live at home, when Jeff and I see her, we observe her extreme fatigue caused by cystinosis. We listen with concern when she tells us that her eyes are swollen and hurt. We have seen her crawl back into bed many times through the years because her stomach aches or because nausea overwhelms her. Too often she has no choice but to rest. As she has grown older, I quietly panic when Natalie says she has trouble swallowing because I fear muscle wasting. When she is home, I frequently catch my breath when I walk into her room and see her nightstand covered with bottles of medication – all the meds that keep her alive. It is a sight I will never get used to. Her routine is certainly not "normal," but I remind myself that it is normal for her and normal for others with cystinosis.

The powerful medications used to treat cystinosis, cause daily and painful side effects. Medications are taken day and night – it is a demanding, never-ending protocol – with no days off. On average, our children take between 8-12 medications every day. For Natalie that equates to 42 pills a day or 1,260 pills a month or more than 15,000 pills a year! Like others with cystinosis, Natalie also takes eye drops several times a day to clear the painful cystine crystals that form on her corneas as a result of corneal cystinosis.

> OUR CHILDREN TEACH US ABOUT LIVING LIFE TO THE FULLEST; THAT NOTHING SHOULD BE TAKEN FOR GRANTED; AND THAT THERE IS JOY IN EVEN THE SMALLEST ACCOMPLISHMENT.

Medical complications increase in severity and number resulting in new and ever-increasing symptoms and treatments. There are unending doctor appointments, G-tube feedings, frequent blood draws, growth hormone shots, kidney transplants, bone pain, daily vomiting, painful bloating, eye pain and severe gastrointestinal side effects. As the disease progresses, their bodies deteriorate. The most severe complications for adults are muscle wasting, myopathy, pulmonary issues and the progression of corneal cystinosis.

Although Natalie cannot escape cystinosis, she will not let it control her and she is certainly not defined by it. It is a terrible disease but having cystinosis does not mean a life of sadness or hopelessness. Natalie continues to live her life to the fullest, with courage, determination – and above all, grace.

Despite the dread of cystinosis, it is also accompanied by many silver linings: our children teach us to live life to the fullest; that nothing should be taken for granted; and that there is joy in even the smallest accomplishment. I truly believe that if cystinosis was not part of our lives, we would not know how to love so deeply, passionately and so fiercely.

Research Gives Us Hope

Although cystinosis creates obstacles, they are not insurmountable. Our strategy is to fund the most brilliant researchers in the world who are committed to finding better treatments and a cure. Research gives us HOPE and hope allows us to live with cystinosis until the day a cure is found.

Since 2003, the Cystinosis Research Foundation (CRF) has become the largest fund provider of cystinosis research in the world – raising more than $33 million. CRF has changed the course of cystinosis by investing donors' gifts strategically and aggressively to create a thriving research community. From the beginning, all CRF operating costs are privately underwritten so that 100 percent of every dollar donated goes to research.

CRF has issued 134 multi-year research studies at leading research institutions in 12 countries. CRF-funded researchers have published 63 articles in prestigious, top-rated journals. Those articles, available to everyone in the world, have added to the understanding of the pathogenesis of cystinosis.

Initially, our strategy was to fund basic and bench research. As discoveries were made in the lab, we funded clinical research, and now we are translating that research into new treatments. One of our earliest milestones, and the one of which we are most proud, is having funded every bench and clinical trial that led to the discovery of a delayed-release form of the life-saving medication cystinosis patients must take. The delayed-release medication, approved by the FDA in 2013, is considered the most significant advancement in the treatment of cystinosis in 30 years. Recently we reached another milestone; we have the first donor stem cell transplantation trial at UCLA, which brings us tantalizingly close to a cure.

Moving forward, we have targeted several areas of research including muscle wasting, neurological issues, corneal cystinosis, and stem cell and gene therapy, all with the goal of finding better treatments and a cure for cystinosis.

CRF Issued $1,581,820 for Bench, Clinical and Translational Research

We are pleased that in 2015 CRF issued nine grants totaling $1,581,820. The new grants bring us that much closer to better treatments and a cure. (See details of the 2015 grants on page 74.)

Leveraged Donations Accelerate Research

CRF has issued many grants that the National Institutes of Health (NIH) or other large granting agencies would typically not fund because of the risk and small patient population. As a testament to the outstanding quality of research, CRF grants have been leveraged during the last five years. Stéphanie Cherqui, PhD, from the University of California, San Diego, and Jennifer Simpson, MD, from the University of California, Irvine, have received more than $7.0 million from the NIH and other granting agencies to expand cystinosis research. Stéphanie's most recent NIH grant, awarded in March, was for more than $1.3 million. The grant will allow her to accelerate her stem cell and gene therapy work on cystinosis. Stéphanie has been awarded over $5.5 million in the last four years. Your support has allowed us to fund researchers whose work exponentially increases the breadth and knowledge about cystinosis.

Nanotechnology and Corneal Cystinosis

We remain committed to finding better treatments for corneal cystinosis. Corneal cystinosis is the build-up of cystine crystals in the eyes that causes photophobia (extreme sensitivity to light), severe eye pain, and sometimes, blindness. There is an existing treatment but it is rigorous, painful for some and requires hourly dosing of medicated eye drops.

Ghanashyam Acharya, PhD, and Jennifer Simpson, MD, have collaborated on a novel treatment for corneal cystinosis. Dr. Acharya invented a nanowafer that we believe will revolutionize the treatment of corneal cystinosis. The nanowafer can be loaded with cysteamine medication to treat corneal cystinosis. It is placed in the eye, and as it dissolves, the medication slowly releases in the eye offering relief for hours. CRF owns the license for the cysteamine-loaded wafer, which allows us to drive the research forward as quickly as possible with the ultimate goal of a new FDA approved treatment for corneal cystinosis. Dr. Acharya has been working day and night to ensure that this potential treatment will reach patients with cystinosis. This is a multi-year project but we have an excellent team in place dedicated to working with us to reach our goal.

Stem Cell and Gene Therapy

Stem cells hold the promise of a cure for cystinosis. Dr. Cherqui's work is our hope – not only has the treatment reversed cystinosis in mice, last year she successfully cured corneal cystinosis in the mouse model.

CRF works directly with Dr. Stéphanie Cherqui, whose focus is stem cells and gene therapy. Dr. Cherqui has worked closely with the FDA for several years and is awaiting FDA approval this year to commence the first autologous stem cell and gene therapy clinical trial. We are optimistic that the FDA will approve this study in 2016. Once it is approved, we will help recruit patients.

Cystinosis Research Helps Others

Many of the discoveries made by CRF researchers have applications to other more prevalent and well-known disorders and diseases. CRF-funded stem cell research will help other corneal diseases, kidney diseases and genetic and systemic diseases similar to cystinosis. Finding a cure for cystinosis will help find cures for other diseases.

We Have Come So Far, But Our Work is Not Done

We are on the brink of new treatments and we are close to the cure – but we are not there yet. There are still studies to fund and clinical trials to support. Clinical trials take time and they are expensive but with your continued help, we will make these new treatments a reality.

We are surrounded by all of you who have embraced our children and our community. You have committed your time, energy and resources to help us find a cure for cystinosis. It has taken each of us, working together for our common goal of finding a cure for cystinosis to ensure the research moves forward. CRF is the only foundation in the world funding millions in new cystinosis research grants every year. We are the lifeline that allows the cystinosis research community to thrive. Without your support, we would not have hope.

IT HAS TAKEN EACH OF US WORKING TOGETHER – FOR OUR COMMON GOAL OF FINDING A CURE FOR CYSTINOSIS – TO ENSURE THE RESEARCH MOVES FORWARD.

Thank You for Being Part of the Cure

Now more than ever, we are in a race to save our children – we cannot be complacent. Every day we pray that Natalie and the other children and adults with cystinosis will have a life free from this horrific disease. We pray for a life without medications, without pain, without muscle weakness, without hospital visits and blood draws, and without worries about life expectancy.

We are extraordinarily thankful and eternally grateful for your support. We are resolute in our desire to save our children. Our faith is strong and our determination steadfast and with your help, there is no doubt that we will find the cure for our children.

Thank you for supporting cystinosis research, for standing by our side and for embracing our community.

With heartfelt thanks,

Nancy and Jeff

HOPE THROUGH COMMUNITY

Natalie's Message of Hope

as shared at the Spring 2016 Natalie's Wish Celebration

Good evening and welcome to the Natalie's Wish event. As many of you already know, I am Natalie Stack and as you also know, I have cystinosis. I turned 25 years old in February and I am at USC attending graduate school. I have been studying social work for the past two years and will be graduating this May with a Master's in Social Work! I am so excited to be graduating but that also means I need to get a job! So if any of you need a qualified social worker in your office … come talk to me later tonight!

Over the past two years I have become my own person. This academic year I have been able to intern at the Office of the Federal Public Defender in Los Angeles and I am taking a class on social work and the law. I have learned an extensive amount of information about the criminal justice system and have developed a strong interest in social justice and criminal justice reform. Who knows, maybe I will be going to law school in the next few years!

In addition to gaining the skills and the knowledge needed to thrive as a social work professional, I have grown at a personal level. I appreciate my life more than ever before. Even though I have a chronic illness, I still am passionate about life and all that it has given me. I have been blessed with an incredible support system; my friends and family have always been there for me, supporting me in every way possible. My mom and the cystinosis community have given me hope that I never thought I would have.

This disease will never be in the way of my journey because I won't let it get in the way. Cystinosis does not make me weak, it is not my enemy. In fact, it has made me stronger as a person and more courageous. It has forced me to never give up on life because life is a miracle in and of itself. Cystinosis challenges me every day to be the best I can be and, though some days are just unbearable, every day I start anew and I think of all the love and happiness in my life.

I want to thank cystinosis for letting me see the beauty in life and for allowing me to see the beauty in every person who is affected by this disease in one way or another. Although this disease can be exhausting, overwhelming and downright depressing, we always have each other to lean on.

I want to thank you for coming here tonight and supporting CRF. A special thank you to the researchers who are here tonight and to each and every one of you who have supported the research that will lead to a cure. I am always astounded by the amount of support and love I see at every Natalie's Wish event.

Thank you and enjoy your evening!

Natalie Stack

Cystinosis is one of the 7,000 rare or "orphan" diseases in the United States that collectively impacts approximately 30 million Americans. With such a rare disease, research money is scarce to nonexistent. Yet research on diseases like cystinosis often leads to advancements in other rare diseases.

CRF-FUNDED RESEARCH IS PROVIDING HOPE!

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

100%
OF THE FUNDS RAISED SUPPORT CYSTINOSIS RESEARCH.

The *Cystinosis Research Foundation* is the largest fund provider of grants for cystinosis research in the world!

SINCE 2003

$33 MILLION has been funded and committed to cystinosis research through the *Cystinosis Research Foundation*

63 ARTICLES PUBLISHED *from CRF-funded research!*

YOUR GENEROSITY HAS FUNDED

134 MULTI-YEAR STUDIES IN **12** COUNTRIES

YOUR COMMITMENT HAS GIVEN NEW HOPE TO

2,000 cystinosis patients throughout the world

1 *FDA-APPROVED* **DRUG**

1 *ALLOGENEIC STEM CELL TRIAL AT UCLA*

1ST GLOBAL INTERNATIONAL PATIENT REGISTRY

First *LICENSE FOR NOVEL DEVICE FOR CORNEAL CYSTINOSIS*

First *CYSTINOSIS FELLOWSHIP PROGRAM*

THANK YOU FOR YOUR CONTINUED SUPPORT

Every patient, family and donor from all over the world has changed the course of cystinosis and has given hope to our community. There was essentially no research prior to 2003, and now look!

CURRENTLY THERE IS NO CURE FOR CYSTINOSIS, BUT THERE IS

Hope!

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

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

Cystinosis affects approximately 500 people, mostly children, in North America, and fewer than 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.

CYSTINOSIS RESEARCH FOUNDATION
RESEARCH • HOPE • CURE

TODAY, CRF IS THE LARGEST PROVIDER OF GRANTS FOR CYSTINOSIS RESEARCH IN THE WORLD.

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 provider of grants for cystinosis research in the world, funding more than 134 studies in 12 countries. CRF has raised $33 million, which it has granted or committed to cystinosis research studies around the world. CRF's efforts have changed the course of cystinosis research 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.

A WHOLE WIDE WORLD OF HOPE

MINDY GLEDHILL

MUSICIAN

SAM AND LARS JENKINS
Salt Lake City, Utah

Mindy Gledhill with Stephen, Lars, Ashton and Sam Jenkins

Mindy Gledhill's joyous music is at the heart of an evening full of grace and special memories

By Dennis Arp

Agreeing to do a benefit concert for young Sam and Lars Jenkins, who both have cystinosis. That was the easy part for Mindy Gledhill. The singer-songwriter was born to perform, and generosity comes just as naturally.

But the mother of three had been taking a break from live performance, and as she readied to launch into one of her most familiar songs, "Whole Wide World," she realized she couldn't remember how it began.

To her rescue came a 12-year-old girl in the front row. The youngster started singing for Gledhill, and in a flash the two were vocalizing together. What could have been a stumble instead became a warm and charming step into an evening of full-hearted harmony.

"It wasn't embarrassing, but really sweet," recalls Gledhill, a popular performer in and around Salt Lake City, Utah, where the Jenkins family lives.

"As a musician, I think you have to trust on stage that the audience will be there with you to share the journey," Gledhill adds. "We're all human, and there's usually not a show I do that something doesn't go wrong. You learn to navigate those moments with grace, and you see that they have the potential to be full of magic. That moment became a favorite of the night."

And the night became a resounding success on a multitude of levels – bringing a hopeful community to its feet, raising awareness of cystinosis and attracting donations of more than $21,000 for the Jenkins' nonprofit organization, Sam's Hope for a Cure, which works in partnership with the Cystinosis Research Foundation.

So many people made the evening possible, from Sam and Lars' grandmother, Leslie Jenkins, who spearheaded the show, to the young boys' Aunt Sarah, who brought her signature cake bites to sell. Other family members contributed cookies, cupcakes and Rice Krispy treats.

> "IT'S SOMETHING I DEFINITELY WANTED TO SAY *YES* TO."

A silent auction at the event and on the Sam's Hope website brought in more contributions, and family friends Rand and Lynette Patterson presented a check for $1,550 from a fundraiser at their business, Rand's Auto Sales.

It all added up to a night that matches the spirit of Gledhill's music and lyrics, capturing "the determination, confidence and hope we need," says Stephen Jenkins, MD, Sam and Lars' dad. "Mindy performed an amazing set."

Doing the benefit show to support Sam, Lars and cystinosis research was a no-brainer for Gledhill.

"When I heard about these two little boys, these brothers, and the challenges they face, it's something I definitely wanted to say yes to," she says.

"We packed it to the brim, and I tried to enjoy every minute of it," the singer adds. "I feel at home performing; it feels more like a sharing experience than a 'watch me' experience. It felt like that in that room. We were sharing this really special moment in time, helping people in our community who need help. It was really beautiful."

Among the standout moments was when she sang "Hourglass," a piece Gledhill wrote for her own three children.

"It's a special lullaby for boys, and performing it was a moment in which I felt particularly strong emotions," she says.

Then there was the impromptu duet that launched Gledhill into "Whole Wide World." The song begins, "I'm gonna walk a hundred miles, I'm gonna whistle all the while, if that's what it takes to make me smile, I'm gonna walk a hundred miles."

And on such a journey, who doesn't need a little help?

"It reminds me of my purpose as a musician," Gledhill says. "To me, it's a very spiritual job – connecting with people's journeys in life. And that experience makes me a better person."

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CYSTINOSIS CHAMPION

THE COLOR OF KINDNESS

Grocer Scott Rowland helps transform the Little family's "Paint the Town Purple" campaign into a display of what it means to be a community

SCOTT ROWLAND

GROCER

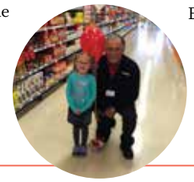
OLIVIA LITTLE
Port Elgin, Ontario, Canada

By Dennis Arp

Like so many Canadian stories, this one centers on warm-hearted people, a tight-knit community and hockey.

Saugeen Shores, Ontario, is a picturesque lakeside town of about 12,000 residents who root for each other and for the Winterhawks, the local senior hockey team. Chad Little used to play for the team, and when his daughter Olivia was diagnosed with cystinosis, the organization naturally jumped in to support the search for a cure.

It was through the president of the Winterhawks that Chad and his wife, Erin, met Scott Rowland, who quickly became the Little family's MVP – most valuable partner in raising funds for the Cystinosis Research Foundation and awareness about the devastating effects of the disease.



Rowland is a grocer who, along with his wife, Kathy, owns and operates an independent store in town. Rowland gets inundated with requests for support of local causes, "so we have to be selective," he says.

But there was plenty in the story of Olivia's fight against cystinosis that caused the Rowlands to make an all-in commitment.

"The Littles shop in the store all the time, and they're lovely people," he says. "When you hear about the things Olivia goes through in a day and what the family faces in providing her care, it draws on your heartstrings. You want to do all you can to help."

Two years ago, the Rowlands' store became a hub of the Littles' "Paint the Town Purple" campaign. Balloons, bows and ribbons in Olivia's favorite color festooned the aisles of the store, and for a week customers rose to meet the opportunity to contribute. The week of generosity netted $5,000.

Things went so well that this past year the Rowlands expanded to two weeks of giving.

"Not just me but the entire staff gets behind it," Scott says. "It's a small community, and to see the whole town support the campaign just shows that being small is not all that bad."

Each of the store's cashiers catches the spirit, buoyed by a friendly contest Rowland launched to reward those who garnered the most $1 or $2 donations to aid the CRF mission. The champion cashier earns a $100 gift certificate, with $50 and $25 going to the second-place and third-place finishers. An auction of Winterhawks game-worn jerseys adds to the fun and the amount raised.

> "AS PARENTS, WE TELL OLIVIA SHE CAN BE ANYTHING IN THE WORLD, AND WHAT SHE SAYS IS, ONE DAY I WANT TO WORK FOR MR. SCOTT."

The whole town embraces the campaign. But no one catches the fever quite like Rowland.

"It's exciting to go in every day to check the computers to see how we're doing, and then share the news with the staff," he says. "When we hit our target, there's a big celebration that brings us all together."

That target was $10,000 this time, which made the celebration twice as sweet when they reached the great moment.

"Scott's a great human being for our whole community," Erin Little says. "The money is wonderful – $10,000 makes a huge difference. But because of Scott and his efforts to be a leader and raise awareness about cystinosis, the impact multiplies."

The campaign culminated with Olivia, 6, and her sister, Harper, 3, joining in a big thank-you day of grocery-bagging for the town's customers.

"There's nothing like a fresh package of bread for a 3-year-old to squeeze," Erin says with a chuckle. "And Olivia is very charming – smiling, happy. She gets to run amok. They absolutely love it.

"As parents, we tell Olivia she can be anything in the world, and what she says is, 'One day I want to work for Mr. Scott.'"

What Erin and her family get is a chance to experience the depth of their community's commitment to making Olivia's life better. They get to meet and know the generous families, like the one that held an impromptu garage sale, then brought in a plastic bag filled with 24 dollars and some-odd change. And the woman who heard about Olivia and cystinosis, then made a special trip to the store to donate $100.

"Scott has done this," Erin says. "It's not just the money – it's the human compassion he's creating. It's a campaign that turns into bake sales and fish fries, and tons of other things that bring our community together – all for a little girl named Olivia and this rare disease.

"It's a display of what it means to be a community. This is what that looks like."

The thing is, it looks like the Rowlands are just getting started. For the next campaign, they're pushing things to three weeks, and the new goal is $15,000.

Who would argue that they won't make it?

"My wife and I are committed to this," Scott says, "and we will continue to be until we find a cure."

A BID FOR THE BETTER

Supporting the Head family's fundraiser, Laurie Lanway rises to meet a last-minute challenge and becomes a champion for life

LAURIE LANWAY

FAMILY FRIEND

By Dennis Arp

MARY HEAD
Rainier, Washington

Every day at the school where she works, Laurie Lanway eases the burden of the special-needs children in her charge. So when she saw her friend from church, Melissa Head, facing her own special needs, Lanway leaped in to make things better.

The occasion was the Head family fundraiser, "Music for Mary," supporting daughter Mary and cystinosis research. Head made all the preparations for an evening of food and fun, but then right before the doors opened a flood of last-minute auction items created a bit of a crisis. Everyone was feeling overwhelmed.

Everyone but Lanway, that is.

"Laurie was wonderful," Head says.

Lanway saw that the 20 or so new auction items got logged in and became a part of the program. Then when the bid sheets started to to run out, she tracked down some notebook paper and started crafting makeshift sheets to fill the gap.

"It did get challenging at the last minute," Lanway acknowledges.

Even as some guests were already bidding, Lanway was creating new sheets so others could join the fun.

"When people are eager to donate, you certainly don't want to turn them away," Lanway says. "Every dollar helps."

For the 200 attendees, the night resounded with warmth and reverie. And for the cause of cystinosis research, the donations reached a robust $19,000.

"It went from one of the most stressful nights to one of the most fun," Head recalls.

In short order, Lanway went from earnest friend looking for ways to help to dedicated champion of the cause – someone who contributes in a multitude of important ways.

She now leads the meetings of the five-person committee that advises and works on Head family fundraising events such as "Music for Mary." But she isn't the kind of person who limits herself to a job description or traditional duties.

"She jumps in and does whatever needs to be done," Head says of Lanway. "This cause is very real and important to her; she reads the literature. She has a heart for serving, and she wants to make a difference."

Much of the difference Lanway makes is in relieving the day-to-day pressures Head faces as a parent of a young cystinosis patient.

"I took the lead role so Melissa wouldn't have to be the (fundraiser), so she can be the mom," Lanway says. "I want her to be able to sit back and fill the important roles she needs to fill."

> "I WANT TO DO WHATEVER I CAN TO HELP MELISSA KNOW THERE'S EVERY REASON TO HAVE HOPE."

Of course, Head fills no bigger role than caring for Mary, who at age 7 has been meeting the many challenges of cystinosis since she was first diagnosed at 9 months. The support of her husband, J.R., and their three other children makes a world of difference for Head, as does Mary's resilience in the face of daily nausea, a daunting regimen of medications and the myriad other obstacles cystinosis concocts.

"She's quite a trooper, " Head says of Mary.

Lanway also feels the family's deep connection to the cause.

"I've known Mary since before she was here," Lanway says. "When she was born, there was such joy, and when the diagnosis came, it broke my heart."

So when Head returned to her home in Ranier, Washington, from the recent Cystinosis Research Foundation Day of Hope Family Conference, Lanway was heartened to hear about advances in treatment and progress toward a cure.

Along with the rich friendships she has developed, such news makes it easy for Lanway to redouble her efforts to support Melissa, Mary and all cystinosis families.

"I couldn't even describe how deep the connection is," Lanway says. "I love that I get to be their champion.

"They inspire us to work toward raising more and more funds, because we see that it's really making a difference. I want to do whatever I can to help Melissa know there's every reason to have hope."

THE HEROES NEXT DOOR

Childhood friends Sara and JJ bring love from the old neighborhood and add new vigor to the Alexanders' "Hearts for Hadley" event

SARA AND JJ ASTORQUIA

FAMILY FRIENDS

HADLEY ALEXANDER
Boise, Idaho

By Dennis Arp

There's something bright and exciting about a new beginning. Then there's the scary part.

When the Alexander family moved from Seattle to Boise, their plans for a fundraiser to support their daughter Hadley and cystinosis research moved with them. If only they had trusted friends already in town to smooth the edges of the transition. If only those friends brought special skills that made everything less daunting. If only they could all work together as if they'd known each other forever.

If only everyone could have friends like Sara and JJ Astorquia.

Those loved ones mean the world to Marcu and Ben Alexander, who consider the Astorquias their champions. Ben grew up in Twin Falls, Idaho, sharing a hometown and childhood friendships with Sara and JJ.

"Ben says that they've been in his life as far back as he can remember," Marcu relates.

"Their family was like my extended family," says Sara, who grew up next door to Ben.

"It really feels like we're brothers and sisters," JJ adds.

So when life made them all neighbors again in Boise, Sara and JJ jumped at the chance to help make the Alexanders' fundraiser, "Hearts for Hadley," a rousing success.

"We knew from the moment Hadley was diagnosed that we wanted to raise money for cystinosis research," Marcu Alexander says. "But it's all a little bit scary. First we sold 'Hearts for Hadley' T-shirts, and then we had a poker night. But it was only after we moved to Boise that we tackled a bigger event."

It was Sara and JJ who helped make that possible.

The plan for the more ambitious "Hearts for Hadley" event included music, food and both a silent and live auction.

"We had been involved with quite a few nonprofit organizations, so we knew a bit about how to run them and succeed with events," Sara says. "We just had a few extra tools to contribute."

Contacts in town helped build community support and attract auction items. JJ had never been an auctioneer before, "but I've never been shy about public speaking, and the connection we share made it easy to get up there," he says.

From Marcu Alexander's perspective, the first event was a home run, even though it started with trepidation.

"You wonder if people will come, or if they come, will they have a good time?" she says. "Our goal was $20,000, and we raised $60,000. I was blown away."

The event continues to grow, and the Astorquias keep making it better. The most recent "Hearts for Hadley" event included a dessert auction, which was Sara's idea. A host of friends contributed homemade goodies that generated friendly bidding wars.

"People would buy a cake for $300 and then share it with the entire table," Marcu says. "We raised way more than we ever expected."

> "THE REWARDS FOR US ARE JUST KNOWING WE CAN BE INVOLVED AND MAKE A DIFFERENCE."

Another fresh idea came courtesy of JJ's friendship with popular newscaster Mark Johnson of KTVB-TV in Boise. Not only did Johnson do a feature story on Hadley and cystinosis that increased interest in the event, but he also co-hosted with JJ.

Then there's the contribution of musician David Andrews, who performed a show to benefit "Hearts for Hadley."

It all adds up to mountains of support for 5-year-old Hadley Alexander, her parents, her sister Stella and the Cystinosis Research Foundation.

"The rewards for us are just knowing we can be involved and make a difference, plus seeing where the money goes – all the changes in medicine and technology," Sara says. "Research is being funded, and that's rewarding in itself."

It's fun to share special moments with lifelong friends, but the ultimate goal remains on the horizon, the Astorquias note.

"We're all hoping we can find a way to get a cure for all of these kids," JJ says.

Teachers and Staff (L-R):

Patrick McIntosh (Principal), Renee Malaki, Pia Glenn, Jane Dolcini, Steve Serain, Penny Blocker, Sharon Wong, Sean Kinlock, Serena Lim, Rosemary Mendoza

DEEP-DISH AFFECTION

Teachers and staff at Holy Spirit School prepare a farm-to-fork feast that shows the Partington family just how much they're loved

HOLY SPIRIT SCHOOL

TEACHERS AND STAFF

JENNA & PATRICK PARTINGTON
Sacramento, California

By Dennis Arp

Sean Kinlock, Teresa Partington, Betsy Reed

As Jenna and Patrick Partington's fourth-grade teacher Betsy Reed saw first-hand the children's daily struggles with cystinosis. She could tell when they were ailing, when a restless night left them lethargic, when their medications made them queasy.

But she also saw them smile broadly, work diligently and make friends easily.

"They never used cystinosis as an excuse," Reed says. "They never complained and they never quit. I admire them greatly."

Admiration runs deep at Holy Spirit Parish School, where the faculty and staff lift up the Partington family and help ease the many burdens of coping with cystinosis.

"Everyone there knows our story, and they're supportive of the kids and us," says Teresa Partington, mother of Jenna and Patrick. "They are taking the journey along with the kids, along with all of us."

Recently that journey took a spirited and tasty turn. Rather than just take up a collection for Jenna & Patrick's Foundation of Hope, staff members at Holy Spirit School decided their contribution could go farther if they combined it with their creative talents. They used the money to buy the food and fixings for a farm-to-fork dinner they prepared themselves. They auctioned the meal at the Partington's Swing & Bling fundraiser in their hometown of Sacramento.

> "THEY ARE TAKING THE JOURNEY ALONG WITH THE KIDS, ALONG WITH ALL OF US."

The whole school community got involved to make the dinner an evening to remember.

Reed is a key organizer and tended bar at the event, where she mixed up two signature cocktails. "We called them 'Holy Spirits,'" she says.

Dona Pollacchi was Jenna and Patrick's kindergarten teacher, and she helped decorate the setting for the outdoor dinner. Jessica Lugo was the chief creative force behind the decorations.

Meanwhile, Renee Malaki, a science teacher at the school, brought professional catering skills to the table and filled the role of chef for the evening. Todd and Jennifer Ferrara provided selections from the Clesi winery to accompany the meal, while Dinny and Ron Thomas made their backyard available for the night. And then there are the winning bidders, Jason Gallelli and his family, who made the whole event possible.

From the Holy Spirit School principal to support staff and other parish families, just about everyone found a way to pitch in.

"We all feel blessed that the Partingtons are in our lives," Pollacchi says. "Those children put us face to face with a way to be supportive of life. We all hope that our efforts will help find a cure."

Reed says it was a blessing for her to help with the dinner after she had previously attended the Cystinosis Research Foundation's Day of Hope Family Conference in Newport Beach, California.

"I was amazed at the community that supports cystinosis families there," she says. "They comfort each other so they all know they're not alone. And when we were thinking about that here at Holy Spirit, we knew that we wanted to be that support system for the Partingtons in Sacramento."

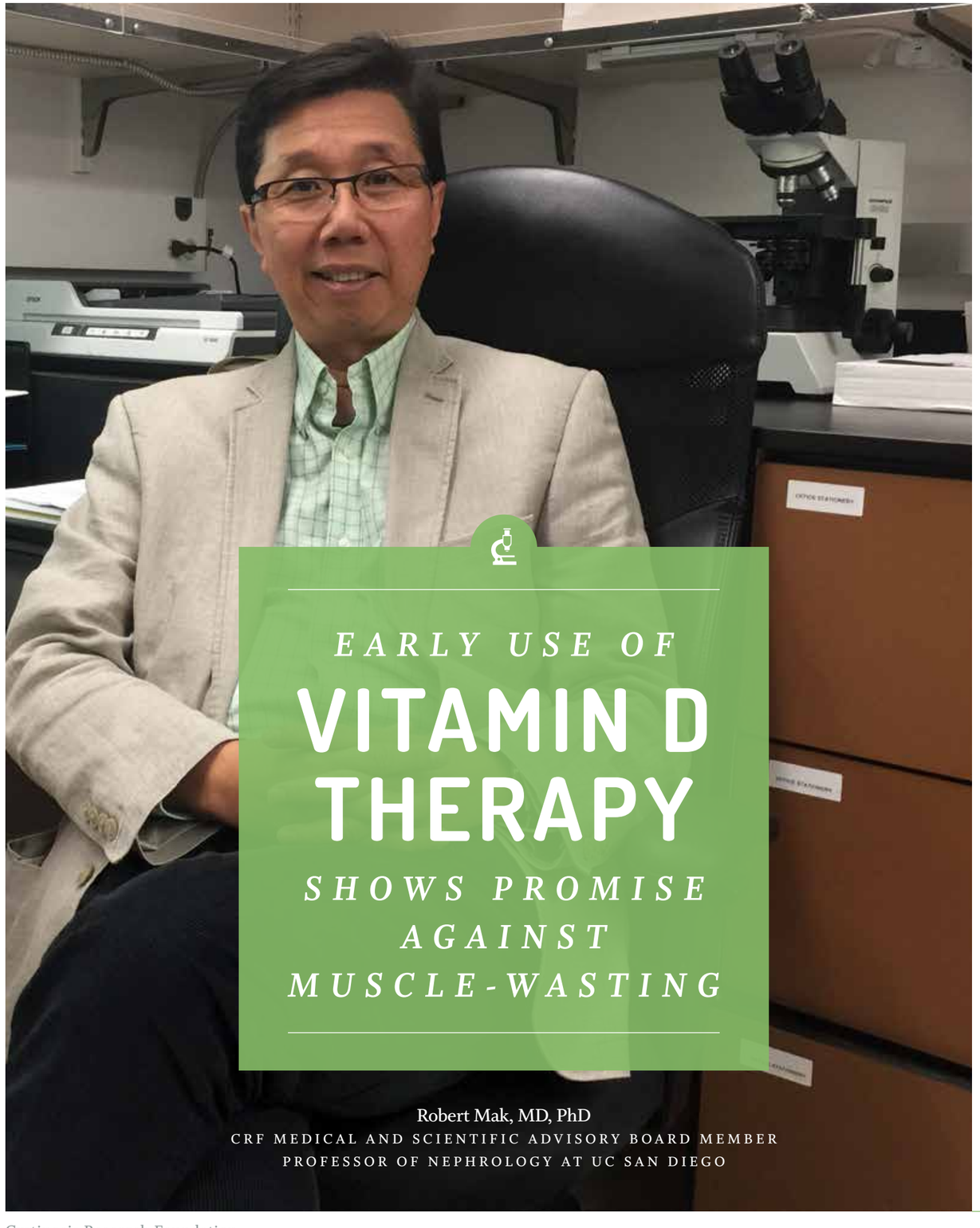
The family feels the love and tries to return that positive spirit every day, Teresa and Kevin Partington say. But Teresa can't help but worry that the burdens of cystinosis weigh on the teachers at Holy Spirit School.

During a parent-teacher conference with Malaki last year, Partington explained that she knows Jenna has trouble sitting still, "and I didn't want her to detract from the learning environment." Malaki quickly brushed off her concerns.

"She told me, 'The kids I asked to help Jenna in class are learning as much or more from the experience as Jenna is," Partington says. "That's the kind of positive attitude everyone has."

And whether it's in the classroom, on the playground or at a farm-to-fork dinner, that support helps keep the Partingtons going.

"It's our way of coping – to share the burden by reaching out to those close to us, knowing that they will help carry us along," Partington says. "It's so cliché to say that it takes a village, but this community of people makes such a difference in our lives. They're kind of like a little island of hope for us."



EARLY USE OF VITAMIN D THERAPY SHOWS PROMISE AGAINST MUSCLE-WASTING

Robert Mak, MD, PhD
CRF MEDICAL AND SCIENTIFIC ADVISORY BOARD MEMBER
PROFESSOR OF NEPHROLOGY AT UC SAN DIEGO

By Dennis Arp

As research expands and new treatments emerge, cystinosis patients gain the prospect of longer, more rewarding lives. But as they age, these patients often face a new set of side effects. One of these is the wasting away of muscle function.

Aided by funding from the Cystinosis Research Foundation (CRF), Dr. Robert Mak studies the energy balance in cystinosis patients and how best to combat muscle-wasting. His research offers the hope of new therapies that may help treat or prevent muscle dysfunction.

In particular, there's growing evidence that vitamin D can play an important role in helping cystinosis patients avoid the worst effects of muscle-wasting. Under the right circumstances, vitamin D supplements can help patients achieve normal models of muscle function, Dr. Mak says.

Even otherwise healthy people can struggle to maintain a balance of nutrition intake and output to sustain proper body mass and muscularity. Meanwhile, cystinosis patients "have two strikes against them," says Mak, MD, PhD, professor of nephrology at the University of California, San Diego.

"Cystinosis patients burn energy at a high rate and have difficulty getting the food intake to match because they have no appetite," Dr. Mak says. "That negative balance leads to a loss of weight and poor muscle function."

As kidney disease progresses over time, that energy imbalance grows and leads to frailty, robbing cystinosis patients of mobility and quality of life. In some advanced cases, muscle-wasting can impede swallowing or even breathing.

Even though many cystinosis patients have few calorie reserves to spend, their bodies often burn brown fat – the kind lean people tend to accumulate – to create body heat that's not needed. Dr. Mak equates this to not making much income but still maintaining a high credit-card bill.

"It's like the body declares bankruptcy," he says.

Research shows that with pathological conditions such as cancer or HIV infection, normal white fat turns to brown. "We're the first to show that this browning of tissue occurs in cystinosis," Dr. Mak says. "This is one of the main causes of muscle-wasting."

To illustrate the extent of the muscle-wasting problem, Dr. Mak cites a recent study that looked at 86 adult patients with cystinosis and found that close to 40 percent had muscle disorders. Treatment with cysteamine does seem to delay onset of muscle-wasting by about five years, he says, but these patients still tend to see deterioration by their mid- to late-20s.

"This is a very serious long-term complication of cystinosis," he notes.

His latest findings show that a vitamin D deficiency happens early in the progression of cystinosis, even before the onset of chronic kidney disease, he says. As the disease progresses, the deficiency can grow, in part because cystinosis patients have to avoid exposure to sunlight, which is a source of vitamin D.

There are two kinds of vitamin D in our bodies. Vitamin 25D circulates in the blood in high concentrations and has a half-life of weeks. But it was thought that this kind of Vitamin D had to pass through the kidneys to convert to the active form, known as 1,25D. So when nephrologists see kidney disease advance in cystinosis patients, they often prescribe 1,25D as a preventive measure.

From his research, Dr. Mak believes patients should be treated with 25D supplements early in the process, before kidney disease presents.

"Now the thinking is that if we can just deliver the raw material to the muscle, the muscle can do the conversion (to active vitamin D)," Dr. Mak says. "We need to look at the problem early and treat it promptly. If we do, I believe it can go a long way to preventing muscle-wasting and neuromuscular complications."

The doctor sees vitamin D supplements as the first line of defense. Supplements need to be carefully prescribed and monitored by a physician, but they're readily available and have few side effects. Dr. Mak hopes to begin a registry of adult cystinosis patients to determine the extent of vitamin D deficiency and build a cohort for a clinical trial of vitamin D supplements.

However for patients with advanced muscle-wasting, "we might need stronger medicine," Dr. Mak notes.

Research using a mouse model shows that cystinosis patients tend to have high levels of leptin, a fat hormone. "If we give a leptin receptor antagonist, we can reverse this problem," Dr. Mak says. "In experiments, we see subjects put on muscle and weight; muscle function is better. That's quite helpful."

But this promising therapy is only in the lab study stage. It is years away from a possible clinical trial. For current treatment of advanced muscle-wasting, "we can possibly think about anakinra," Dr. Mak says.

Anakinra is used to reduce inflammatory response, especially in patients with rheumatoid arthritis. With cystinosis, "we also notice a lot of inflammation," Dr. Mak says.

However, the drug can have strong side effects, including intestinal distress, liver problems and a compromised immune system. Such complications are among the reasons Dr. Mak is putting much of his own energy into exploring the use of vitamin D supplements early on, before the onset of chronic kidney disease.

"We're very excited about this," he says. "We hope to identify more patients who might benefit from this therapy."

2016 INTERNATIONAL CYSTINOSIS RESEARCH SYMPOSIUM

Researchers (L-R)
Julie R. Ingelfinger, MD • Paul C. Grimm, MD • Stéphanie Cherqui, PhD • Pierre J. Courtoy, MD, PhD
Corrine Antignac, MD, PhD • Francesco Emma, MD • William Smoyer, MD, FASN

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

I recently had the privilege of attending the CRF Fifth International Cystinosis Research Symposium. It's a research meeting held every other year where all the scientists who have received funding from the Cystinosis Research Foundation come together to share their findings. It's an opportunity for brilliant minds to collaborate toward our ultimate goals of finding better treatments and a cure for cystinosis.

There were a few things that struck me as I listened to the many research presentations. The first is that cystinosis is a COMPLICATED disease. The classical teaching of cystinosis is that loss of the transporter protein "cystinosin" leads to accumulation of cystine in lysosomes, which subsequently leads to cell death and tissue damage. This mechanism is the basis for treatment with cysteamine, which depletes cystine from the lysosome. If this was the only mechanism by which cystinosis caused disease, then cysteamine should have essentially been a cure. But study after study has shown that even with cysteamine therapy, patients progress to develop Fanconi syndrome, renal failure, thyroid disease and muscle-wasting, with its host of complications. The CRF has funded numerous basic science studies that have taken a closer look at the biological pathways that are disrupted by the loss of the cystinosin protein. It turns out the cystinosin protein does a lot more than move cystine out of the lysosome.

One common theme at the symposium was the role of cystinosin in a cellular process called "autophagy." Autophagy literally means "to eat oneself," and it is an essential process through which cells recycle damaged proteins and organelles to generate nutrients during periods of starvation or stress. Several scientists showed how this process is impaired in cystinosis. Another important cellular process affected by the loss of cystinosin is the mTORC1 pathway, which is involved in sensing nutrient availability and regulating cell growth. Researchers showed that impairment of this pathway has many negative downstream effects in cystinosis and may explain the development of Fanconi syndrome. Yet another important pattern highlighted by researchers was the presence of increased inflammation in cystinosis cells, which may be implicated in muscle-wasting and kidney disease.

The next thing that struck me was how important this basic science research is to finding new therapies. Sometimes it is hard for me to see how studying cystinosis in fruit flies or yeast will lead to a new drug. But by studying these complex cellular pathways, scientists have discovered multiple novel drug targets. For example, Dr. Sergio Catz and Dr. Francesco Emma are looking at experimental drugs that could improve the autophagy pathway in cystinosis cells. Dr. Robert Mak has identified multiple inflammatory cytokines that are elevated in cystinosis, and he is studying the effects of FDA-approved drugs that target these cytokines on muscle-wasting and renal function. Other researchers are looking at the effects of certain drugs on the mTORC1 pathway. None of these possible therapies would've been discovered without first taking a closer look at the cellular level.

Finally, what struck me the most is that there are some very exciting things on the horizon. Dr. Stéphanie Cherqui presented an update on stem cell transplantation. Her lab is working tirelessly to improve the process by which the correct cystinosin gene is inserted into a patient's stem cells. She is on track to submit the application for a human trial with the FDA, hopefully in the fall of 2016. Dr. Ghanashyam Acharya mystified the audience with his nanowafer research, which promises to be an extremely effective tool in treating corneal cystinosis. He predicted starting the human trial by the end of the year. Dr. Francesco Emma has screened 1,200 drugs and found seven new potential candidates for treating cystinosis. One of these drugs may have a better side effect profile than cysteamine, while another drug may act on impaired cell pathways that are not treated by cysteamine. His lab is moving forward in testing these drugs in knockout mice.

The symposium was an amazing example of research in action. A diagnosis of cystinosis is devastating, but we are fortunate to have such a vibrant community of researchers who are dedicated to finding a cure and better therapies. I want to thank all the scientists, physicians and researchers for their diligent efforts. I also want to thank the many donors whose money has created a steady pipeline of funding so that we can always move forward with research. I especially want to thank Nancy and Jeff Stack without whom none of this would have been possible. Every rare disease needs a CRF and a Nancy Stack.

CYSTINOSIS COMMUNITY

THANKS TO OUR DONORS FROM AROUND THE WORLD!

CRF REACHED A RECORD $3.5 MILLION FOR CYSTINOSIS RESEARCH IN 2015

Wear Your Rare Campaign
BARTKOWSKI, MANZ AND MOORE FAMILIES
$1,913

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OLIVIA LITTLE
$71,185

Amanda's Hope for a Cure
AMANDA KUEPFER
$5,752

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Mulligan's Fore Morgan
MORGAN PEACHMAN
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24 Hours for Hank
HENRY STURGIS
$50,000

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$1,145

Tanner's Time for a Cure
TANNER EDWARDS
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CALEB GOWEN
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TINA FLERCHINGER
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Pies and Plays
ALIYAH AND MADELYN WALKER
$10,406

Gabbie's Wish
GABRIELLE STRAUSS
$51,075

Joshua's Journey of Hope
JOSHUA CLARKE
$18,000

A Cure for Keegan
KEEGAN MANZ
$7,233

Jenna & Patrick's Foundation of Hope
JENNA AND PATRICK PARTINGTON
$101,000

Chance for Chase
CHASE CHODAKOWSKY
$27,823

Hope for Holt
HOLT GRIER
$1,750

Music for Mary
MARY HEAD
$14,896

Abbi's Road to a Cure
ABBI MONAGHAN
$2,998

Hearts for Hadley
HADLEY ALEXANDER
$81,728

Bailey Believes
BAILEY DEDIO
$6,481

Race for Nicole
NICOLE HALL
$16,619

Lots of Love for Landon
LANDON HARTZ
$34,180

Sam's Hope for a Cure
SAM AND LARS JENKINS
$18,230

Hopes & Wishes
JAKE KRAHE
$16,005

NATALIE'S WISH CELEBRATION 2016

THIS IS OUR FIGHT FOR A CURE!

A HOPE-FILLED EVENING TO REMEMBER

IT WAS A NIGHT TO REMEMBER.

On April 9, 485 people joined together from around the world for the cystinosis community's largest fundraiser, the Natalie's Wish Celebration held at The Island Hotel in Newport Beach, California. It was a truly inspiring and hope-filled evening, raising a record-breaking $3.3 million for cystinosis research.

Attendees traveled from across the globe to celebrate the milestones we have reached, the discoveries we have funded and focus on the incredible work still ahead. Guests included 55 cystinosis families and adults with cystinosis from as far away as Canada and Sweden. United in the quest for a cure, many of the families raise funds for cystinosis research in their own communities. It is always a highlight when the families proudly parade on stage to present their checks. The gala was the finale to the three-day Day of Hope family conference.

Guests entered a dazzling and enchanting ballroom decorated with a message that set the tone for an inspiring evening of unparalleled success. This is Our Fight For a Cure. >

Rachel Platten's performance moved the crowd with her inspirational songs.

The celebration featured a performance by the charismatic and inspiring singer songwriter Rachel Platten who performed her blockbuster hit, "Fight Song," her new release, "Stand By You," and many other songs that had the audience singing along with every word. The grand finale of Rachel's show was a guest performance of "Fight Song" by none other than 24 children from the cystinosis community. It was a touching moment that had a moved crowd up on their feet for a standing ovation.

Another special part of the evening was the live auction that raised $330,550. The auction featured eleven items including fine dining, luxurious getaways and extraordinary wine collections.

The night was filled with moving stories from many members of the cystinosis community. This year, Natalie Stack shared where she is now: about to graduate from USC with a master's degree in Social Work. Natalie beamed as she thanked members of the community for their continuous support. Following Natalie, her mother, Nancy Stack, spoke of the optimism that donors and researchers have provided her family and the cystinosis community year after year. Later in the evening, Lauren Hartz, mother of Landon, age 6, with cystinosis, shared her family's journey through treatment. She spoke of the ups and downs, and of her family's immense gratitude for the Cystinosis Research Foundation and the Stack family. Each speaker's story was filled with hope and was nothing short of inspirational.

The Stack family would like to thank all who attended and each and every member of the community who has helped provide every child living with cystinosis the greatest gift of all: the blessing of hope.

LAUREN HARTZ'S SPEECH AT NATALIE'S WISH CELEBRATION

INVENTORS & MAGICIANS & HOPE

One of the most touching moments of the 2016 Natalie's Wish Celebration was a speech made by Lauren Hartz, mother of Landon, with cystinosis. Here are the heartwarming and inspiring sentiments she shared that evening:

To say that I am honored to stand here in front of you today would be an understatement. My name is Lauren Hartz and my first son, Landon, was diagnosed with cystinosis when he was 14 months old. I wish I could say that was the beginning of our journey, but it began well before that.

I have memories of sitting on the kitchen floor encouraging him to take a bite of something, anything. My husband, Jimmy, used to call me from work to ask how we were doing and what Landon ate. He was losing weight, malnourished and diagnosed with failure to thrive. I felt like a failure. We took test after test to find the problem, and continued to get the same answer. I was told that there was no medical reason for Ladon's lack of appetite and that we needed to admit him to Children's Hospital of Pittsburgh for intense feeding therapy.

When we received instructions for admission, I was told that they were going to collect another urine sample. The doctors had found sugar in his urine, which was probably a fluke, but they needed to be sure. Soon after the sample was taken, the nephrology team presented, like a small army, in our room. Within hours of being admitted, we learned a whole new vocabulary … Fanconi syndrome, cystinosis, transplant… It felt like a nightmare.

The doctors told us not to Google cystinosis. Jimmy made me promise not to, but my mind was thinking the worst. I did, and that's when we came across the Cystinosis Research Foundation's website and I reached out to Nancy. She quickly responded to my e-mail. Cystinosis wasn't yet confirmed and we already trusted her. She comforted us and shared the amazing research that was being done and the fact that we were in this together.

After the diagnosis was confirmed, Jimmy and I looked into how to raise funds for research that would help find more effective treatment and a cure. We had to do something. We felt so helpless. Five months after Landon was admitted to the hospital, with the help of our family, friends and other community members, we

>

Lauren Hartz's speech (Cont'd)

held our first fundraiser, a Halloween party. It was so empowering. Since then, we have held nine fundraisers and have raised over $160,000 for cystinosis research.

I don't know that I can go so far as to call cystinosis a blessing, but this journey certainly has been. There are moments that are hard. It is heartbreaking to watch Landon try to keep up with his peers when he plays sports, see him become lethargic after a busy day, hear someone comments on the smell that is caused by the medicine that keeps him healthy, see his Mic-Key button cause him discomfort, hold him tight as he screams and cries during regular blood draws, and each time we lay him on our laps to give him eye drops. Each normal childhood illness puts us on high alert because we fear he will become dehydrated and end up in the hospital. To be honest, I'm traumatized from our month-long hospital stay. Anytime I think about going back, my heart races and my eyes fill with tears.

However, I would like to share the blessings that have made our journey so special… the Stack family, the cystinosis community whom we have come to love and I consider my best friends, our family and friends who work tirelessly to organize each fundraiser, the goodness and generosity we witness each day and the lessons we have learned about the importance of gratitude. Those are just few.

Recently I had to change Landon's Mic-Key button, which is always challenging. He screams and cries and begs me not to do it. Afterwards, I held my crying boy tightly whispering that I am sorry. He looked at me and asked if I know any inventors or magicians. Landon has an enormous imagination so I wasn't sure what he meant. He told me that he needs to find an inventor or a magician to help make his belly not hurt anymore. I felt tears fill my eyes and then found myself saying, "Buddy, I know a lot of inventors and magicians who are working hard to make sure that your belly stops hurting. They are the people that we will see in California."

To all who support cystinosis research, I am thankful. To those who raise awareness, I am thankful. For those who dedicate their careers to finding better treatments and a cure, I am thankful. And to all of you, who have made our cause your cause, I am forever thankful. Thank you.

"Buddy, I know a lot of inventors and magicians who are working hard to make sure that your belly stops hurting. They are the people that we will see in California."

THANK YOU

FOR JOINING OUR FIGHT FOR A CURE

WITH YOUR HELP, ON APRIL 9, WE RAISED MORE THAN $3.3 MILLION FOR CYSTINOSIS RESEARCH

With the support of everyone who attended the 2016 Natalie's Wish Celebration we are moving ever-closer to making Natalie's wish a reality — *"To have my disease go away forever."* We still have much to do but your ongoing generosity gives real hope to the children and young adults in the cystinosis community who desperately need our help.

$500,000 and above

Anonymous

Nancy and Geoffrey Stack

$250,000 – $499,000

Traci and Thomas Gendron

Jenna and Patrick's Foundation of Hope, *in honor of Jenna and Patrick Partington;* Teresa and Kevin Partington

24 Hours for Hank, *in honor of Henry Sturgis;* Tricia and Brian Sturgis

$100,000 – $249,000

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Laura Khouri and Michael K. Hayde

Liv-a-Little Foundation, *in honor of Olivia Little;* Erin and Chad Little

Susan and Thomas F. Moran

Tina's Hope for a Cure, *in honor of Tina Flerchinger;* Denice and Mark Flerchinger

$50,000 – $99,000

Anonymous

Gabbie's Wish, *in honor of Gabbie Strauss;* Jody and Trevor Strauss

Hearts for Hadley, *in honor of Hadley Alexander;* Marcu and Ben Alexander

Thormahlen Family Donor Fund, Chris and Bill Thormahlen

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$20,000 – $24,999

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Sam & Lars Hope for a Cure, *in honor of Sam and Lars Jenkins;* Ashton and Dr. Stephen Jenkins

$15,000 – $19,999

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Mr. and Mrs. Chris Dialynas

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Hopes & Wishes, *in honor of Jake Krahe;* Amy and Jeremy Krahe

Music for Mary, *in honor of Mary Head;* Melissa and JR Head

Race for Nicole, *in honor of Nicole Hall;* Stephanie and Aaron Hall

THANK YOU FOR YOUR GENEROSITY AND FOR PROVIDING *HOPE*

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Angela and Bill Montgomery
Natalie and Thomas P. Moran
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Michelle and Reggie DelPonte
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Lori and Richard Glucksman
Leigh Ann and Rick Groux
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Donald F. Brown, Linda C. Brown and Brandi Reed, *in honor of Jordan Darling*
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Magaly Rodriguez
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Susie and Bo Burch
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INSPIRATION FROM RACHEL PLATTEN

I MIGHT ONLY HAVE ONE MATCH, BUT I CAN MAKE AN EXPLOSION

This year's 2016 Natalie's Wish Celebration featured an amazing performance by American singer songwriter sensation Rachel Platten. Her blockbuster hit "Fight Song" has resonated around the globe as one of the most inspirational anthems of 2015. Rachel performed hits from her album, "Wildfire" including "Stand By You" and "Better Place" — each with a moving message of love, support and courage. Her last song of the evening featured very special guest performers: 24 children from the cystinosis community. The children joined Rachel in an encore performance of "Fight Song." The touching grand finale had every audience member up on their feet for a standing ovation.

"It really hit my heart to see how connected and supportive the cystinosis families are," shared Rachel after her performance. "It is absolutely beautiful because everyone is suffering but everyone is fighting together."

Rachel wrote "Fight Song" during a time of her life where she needed a reminder to believe in herself. She wanted to share her story of coming face-to-face with numerous rejections, and despite it all, believing in that tiny voice in her heart that wouldn't give up. Her words have moved individuals and organizations all over the world to keep fighting.

"To see people using my words to remember their own strength within is truly amazing," said Rachel.

Prior to her performance, Rachel sat down with Nancy, Jeff and Natalie Stack and heard more of Natalie's journey with cystinosis and the hope the Stack family has brought to the community.

"I loved getting to spend time with the Stack family and hearing their story," said Rachel. "They are such inspiring people."

Rachel's performance and lyrics left the cystinosis community inspired to continue their fight for a cure, reminding each community member to never give up on that little voice of hope.

FIGHT SONG

Like a small boat
On the ocean
Sending big waves
Into motion
Like how a single word
Can make a heart open
I might only have one match
But I can make an explosion
And all those things I didn't say
Wrecking balls inside my brain
I will scream them loud tonight
Can you hear my voice this time?

This is my fight song
Take back my life song
Prove I'm alright song
My power's turned on
Starting right now
I'll be strong
I'll play my fight song
And I don't really care if
nobody else believes
'Cause I've still got a lot of
fight left in me.

CYSTINOSIS COMMUNITY

How Video Strengthened Our Circle of Hope

By Kristen Murray & Nathan deBruyn

ALBERTA, CANADA

It was a passing comment that gave us the idea. A fleeting conversation with a colleague, imbedded with a concerned query about Seth. "Hey, are you guys still giving Seth all of that medication that you were giving him a while back? Does he still need it?"

It was an innocent enough question, but one that provoked much thought and had far-reaching ramifications.

Almost two years since Seth's diagnosis with cystinosis, the disease remains a prominent feature of our lives, a lens through which every aspect of our reality is filtered. Our routine is one of juggling appointments with specialists, visits to the pharmacy and preparation of the 126 doses of medication that Seth takes each week. Each day we fend off thoughts of cysteine accumulation and the possible complications and suffering that our precious boy may experience. Managing round-the-clock administration of the unpalatable concoction of vitamins that Seth takes and striving to normalize it all in the wake of our stress and exhaustion is just what we do.

So integral is cystinosis to our reality, that I was taken aback by my colleague's passing comment. How could something that we live and breathe every moment not be evident to others?

Nathan's photography has always helped us capture what it's like to live with cystinosis with friends and family. In capturing both joyous and tender moments, his work has moved many people to deeper understanding of our lives. And so, if a picture is worth a thousand words…what about a video?

We sequestered ourselves for a weekend, and thanks to the adorable subject matter we had in footage of Leif and Seth, extensive iMovie tutorials, and Nathan's creativity and persistence, we came up with a visual portrayal of our life.

We sent our video, *Living Life…With Cystinosis*, out to friends and family on the eve of our Circle of Hope and began to receive responses within hours. Many expressed how they were touched by the video's images of Seth's medication and shaken to see the mountain of syringes that awaits him each week. Even the boys' nanny, who takes part daily in giving Seth his vites, was shocked to see the huge volume of medication that such a wee body must contend with.

Friends and family told us that the video helped them better understand our reality with cystinosis. They gained a new appreciation of the enormous impact of the disease on our family from moment to moment. They also were grateful to have such a tangible story to share with their friends and families, evoking even more understanding and support. And indeed, as they shared, friends of friends and even strangers from Swift Current, Saskatchewan to Mexico City to Norway to Bhutan wrote to express that they were awed and inspired by Seth's courage and strength and by our determination as a family to live a rich and joyous life in the face of adversity.

Months have now passed since we shared our video. We have been bolstered by the outpouring of love and support that has followed, and are moved and relieved when people ask questions like, "How are things going with Seth's meds? How are you all managing? Is there anything we can do to help?"

We have been humbled by financial support, and are pleased that our video helped to inspire almost $17,000 in donations to the CRF in Seth's honor. We are moved by this generosity and grateful to be able to extend support to CRF research.

We watch our video from time to time, and especially on the darker days, use it as a means to rekindle our strength. For just as we created a video focused on positivity and hope, we must create a life focused on the many gifts that we have and to live life to its very fullest… with cystinosis.

OUR ROUTINE IS ONE OF JUGGLING APPOINTMENTS WITH SPECIALISTS, VISITS TO THE PHARMACY AND PREPARATION OF THE 126 DOSES OF MEDICATION THAT SETH TAKES EACH WEEK.

Hadley's Hopeful Heart

By Maryjo Baker, Hadley's grandmother
BOISE, IDAHO

Being a grandmother is one of the best parts of my life. My husband and I moved to Boise 15 months ago so we could spend time with our daughter, Marcu, son-in-law Ben and their girls Stella and Hadley. Watching my daughter with her girls gives me great joy. Ben and Marcu are incredible parents who keep the lives of their daughters full of stimulating and healthy adventures.

When Hadley entered the family, all four grandparents and Stella were at the hospital. Watching a nurse clean Hadley up, I was shocked at how bright red Hadley was. I've always wondered since her diagnosis at 18 months if that is typical of babies born with cystinosis. The color soon faded away and Hadley appeared to be a healthy child. But soon her insatiable thirst, too often wet diapers and gradual failure to thrive kicked Marcu's mother instincts into high gear and after many doctor visits, the heartbreaking diagnosis was presented. Thank God for the cystinosis family who immediately reached out to Ben and Marcu with support, love and critical information to help them adjust to life with a cystinotic child. With the proper medicine, great medical care and a loving family, Hadley is thriving.

Hadley possesses great curiosity about her world. Grandpa takes Hadley to ZooBoise once a week for a class to learn about animals. There she has discovered a fondness for all sorts of creatures, even snakes! Upon returning from the zoo Hadley rattles off facts about whatever animal explored that day and proudly shows off a craft project related to the critter. Hadley's room is full of items related to her interests and demonstrates her personality. Hadley's sense of humor is rich and she is starting to tell silly jokes. She loves baby dolls, all of whom she has named, including Mimi, Kiki and Isabella. Hadley likes to nestle them in her arms or tuck them into beds, gently crooning lullabies.

Yes, a horrible disease is also a part of Hadley's life, but it does not define her. Most days she tolerates her medications quite well, unless the Cystagon® has been increased, then her tummy hurts and she doesn't want to eat.

Hadley's favorite playmate is her sister Stella. Stella is bright and understands Hadley's illness quite well for a 6-year-old. When Hadley catches a cold, Stella worries her sister may end up in the hospital, a situation that has occurred too frequently in Hadley's short life. Sometimes, Stella helps give Hadley her meds through her G-tube. But most often, the two of them can be found dancing, singing and playing games they make up, filling the house with frequent laughter. They also like watching cartoons, riding their bikes and playing with their dog Sherman and cat Otto.

Hadley offers lots of help around the home. She folds laundry, uses a hand vacuum and dusts like a pro. Stella isn't as interested in chores, but she sometimes gets in on the baking. Both girls love to paint and draw and play with modeling clay. It makes a bit of a mess, but they do know how to pick up after themselves with some encouragement.

When I watch Hadley and Stella play, I sometimes wonder if Stella will donate a kidney to her sister if/when needed. I imagine them growing up together and experiencing school events, sports, boyfriends, sleepovers — all the things I watched their mom do. I also think about what Hadley may go through because of the disease, and how Stella will help her cope. I pray Hadley will always be compliant with her meds, but Stella will be a huge part of that happening as will her parents.

Ben and Marcu with the help of friends and family sponsor a Hearts for Hadley event to help raise funds for the Cystinosis Research Foundation. The evening raises money for the dedicated researchers who seek improved treatments and hopefully a cure. Businesses and friends donate hundreds of items for a silent auction, including a handmade quilt I provide. Marcu speaks to the attendees about Hadley and her disease, moving most in the crowd to tears, then a wild live auction cheers the crowd as often frenzied bidding generates large sums.

We need a cure for this illness, so all the people with cystinosis can live their lives free of the issues cystinosis causes. Hadley talks about being a mommy and a phlebotomist when she grows up. Let's work together to make her dream and all the dreams of people living with cystinosis possible.

HADLEY'S FAVORITE PLAYMATE IS HER SISTER STELLA. STELLA IS BRIGHT AND UNDERSTANDS HADLEY'S ILLNESS QUITE WELL FOR A 6-YEAR-OLD.

Stella and Hadley Alexander

CYSTINOSIS COMMUNITY

WILLIAM MICHAEL MONAGHAN: *the Younger, Bigger Brother*

By Katie Monaghan, William and Abbi's mom
ST. CATHARINE'S, ONTARIO, CANADA

William Monaghan is 7 years old. He doesn't have cystinosis, and yet he has grown up with it. He is the "younger, but bigger" brother to Abbi Monaghan, who is 10 years old and living with cystinosis. When William was told that we would be writing an article about his point of view, he was thrilled. We asked him a few interview questions, and his insights and responses are included here.

Much like his sister, William doesn't know a world without cystinosis. From the moment he was born, it has affected his life. Shortly after his birth, blood tests were done to determine if he had cystinosis. At only 2 months old, William was left in the care of his loving grandparents while his big sister was admitted to the hospital for a week…his mom and dad had no choice but to be at her bedside in the hospital. Since then, there have been a few more surgeries where William has stayed behind with his grandparents. During these times, even at a very young age, he has always worried for his sister. He has seen his sister take innumerable pills and potions as part of her daily routine. William doesn't like that Abbi has to take medication all of the time, "especially because she doesn't like it."

Abbi's brother, William.

Over the years, he has grown to stand strong and steadfast, near his sister's side, but out of the limelight. William has seen Abbi receive multiple gifts, which are not given to both of them. He has seen her publicly recognized several times for her bravery, at school and at community events, for her everyday fight with a rare disease. William and Abbi joke together that she has become "famous" with all of the publicity. William has been dragged along to countless appointments that were not booked for him. He knows that his sister is allowed to eat whatever she wants, whenever she wants, and everyone is thrilled, simply that she is eating…but William does not have the same liberties. It would be hard for any young child to truly understand these clear inequalities and different restrictions. With all this, as parents, we have had many concerns: is he getting enough attention? Does he truly know how much he is loved, treasured and adored?

We can say that our children, Abbi and William, are our whole world, and we love them both endlessly and unconditionally. However, William came along in some of our darkest days, and brought such a vast and shining light. He is truly our miracle. When he was born, he brought a whole new joy to our lives. He is a unique little guy – growing every day! – who has a creative and inquisitive mind. He is curious, sweet, outgoing, loveable, brave, sensitive, strong, empathetic…and every other possible quality that is good. William Michael Monaghan makes us proud to be his parents each and every day.

One would think that, at his age, he would get jealous of Abbi, and the gifts that she receives after difficult appointments or procedures. But, there is no hiding the fact that he is a very proud brother. He is her No. 1 fan! One might think that he would become envious when we host fundraisers, as the main focus is "once again" on his sister. But William is always beaming with pride and bursting with excitement at the prospect of spreading awareness for cystinosis! William always wears his awareness bracelets and T-shirts with pride, and will explain what cystinosis is, why his sister wears sunglasses, or why he is bigger, to anyone that will listen. He is a marketing manager in the making and he speaks passionately from the heart.

As a family, we do a lot of fundraising for cystinosis. It is a major part of our lives. But it isn't our entire life! William enjoys swimming, biking, hiking and snuggling up to watch movies. He is already looking forward to the summer, when he can get back out on his dad's boat for their weekly fishing adventures. He has big dreams of being an engineer when he grows up, just like his dad. William wants Abbi to realize her dreams, too. At this point, he wants her to be "the chef of a potato truck and have some kids, even though she says that she doesn't want kids, just cats."

Yes, Abbi has cystinosis, but we all are living with it. William is the best brother that anyone could ever hope for. He is, at heart, his sister's strongest supporter. Abbi and William are not only siblings, they are partners in crime. We work hard as a family, and as a team, to spread awareness and raise funds for research. What does William hope for in terms of cystinosis? "I hope they just hurry up and find a cure!" We wholeheartedly agree. It is our family's dream that the cure will come, and that it will come in time for Abbi.

William and Abbi Monaghan

Ultimately, we are a family of four. We are a family who enjoys spending time together. We are a family who loves to laugh… a lot! We dream, we explore, and we grow. Our love for one another is endless. The best advice we can give any family is that all children are different, so treat them differently. But love them the same and find your balance. William's selfless nature and enthusiastic approach to life helps us keep the big picture in mind.

Anything else you want to add William?

"I love my family! Even my stinky dog."

CYSTINOSIS HAS CHANGED OUR LIVES FOREVER, BUT IT WILL NOT DEFINE MORGAN.

The Power of Perseverance

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

I am the mother of two beautiful daughters, Morgan, age 8 (with cystinosis) and Madison, age 5 (without cystinosis). As a professional woman, I hope to instill the same principles and values of integrity, loyalty, perseverance and determination in my children as well as inspire them to believe that anything is possible.

Currently in third grade, Morgan is truly thriving medically, academically and socially. But it hasn't always been easy.

When Morgan experienced situational anxiety due to bullying and a medical emergency at school, we sought additional counseling and help. The school pushed for an Individualized Education Program after reviewing Morgan's test scores. The teachers warned that they thought it would be nearly impossible for her to pass standardized tests based on her initial assessment at the beginning of the school year. However, we wanted to encourage Morgan to catch up organically, without an IEP.

We formulated an action plan that held ourselves, and the school accountable. At home, Morgan worked every night for at least 30 minutes, on top of her regular homework, in hopes that we could increase her test scores. We believed in Morgan, and wanted her to believe in herself as well.

When we planned our first golf fundraiser for cystinosis, Morgan was involved in every step. She became more comfortable talking about the disease, and was excited to share our fundraising efforts at school. She was also enjoying the support of all of her friends and peers.

Fundraising not only brought Morgan closer to her peers at school, it brought her closer to other children living with cystinosis. By supporting the fundraising efforts of other cystinosis families, Morgan learned that she wasn't alone in her daily struggles living with cystinosis. These families, who were once strangers, became our "cystinosis family." Their children, Morgan's new friends, make the disease feel a little less rare and offer her a sense of belonging.

Later that year, at Morgan's midyear intervention meeting at school, I listened with tears streaming down my face as the teachers and principal announced that Morgan had surpassed all expectations and had passed both assessments. She no longer required intervention classes during school. The principal noted the complete 360 turn around that he witnessed in Morgan. She had her "swagger back" and was exuding confidence in every step.

Cystinosis has changed our lives forever, but it will not define Morgan. We will encourage her to work hard, show determination, never give up and know that she is never alone. Because, with enough strength of character, anything is possible.

KENZIE'S STORY

By Shawn and Katie Lawatsch, Kenzie's parents

MARINETTE, WISCONSIN

Kenzie was born August 19, 2011, weighing 5 pounds 12 ounces and was 19 inches long. From the beginning, she was on the smaller size but over the course of her first six months, we became concerned about her poor eating habits, constant vomiting, inability to sleep and the fact that she was not growing. The doctors thought it might be colic, so we tried every formula under the sun but she did not improve.

At Kenzie's nine month checkup the local doctor noted that her legs were bowed and her growth was poor. We added supplements to her formula, however, she did not respond. Kenzie was sent to Children's Hospital in Milwaukee with a diagnosis of "failure to thrive."

In August of 2012 when Kenzie was one year old she was diagnosed with hypophosphatemic rickets. After seeing many doctors and trips to Milwaukee for treatment, she seemed to be responding but was not gaining weight. A year later and not much progress, Kenzie was referred to a renal specialist at Children's. On December 12, 2013, after multiple blood panels and urine tests, she was diagnosed with Type II Renal Tubular Acidosis. Kenzie was admitted to Children's that day for further testing. A genetics doctor was called in to determine the underlying cause.

On December 20th, 2013, we had a diagnosis: CYSTINOSIS. It was the most devastating day in our lives. We immediately started to research cystinosis. Children's Hospital and the Cystinosis Research Foundation helped our family understand the treatment that was needed.

We started Cystagon® January 2014, and began to realize the difficulty of treating this disease. The fact that we had to give Cystagon® every 6 hours was the easy part compared to getting Kenzie to take this horrible smelling and tasting medication – it was heart breaking. By January 14th, we were back at Children's Hospital with a lifeless little girl. Her electrolytes were so low and kidney values so high there was literally no life in her little body. The Cystagon® was stopped and after 3 days of intensive care Kenzie was responding well. The silver lining to this set back was on January 17th, Kenzie had a G-tube put in which helped immensely. On January 19th, Kenzie was released from the hospital and back to her silly normal self.

It took a few months of slowly increasing the Cystagon® dose to build her tolerance to the drug. That year we attended the CRF Day of Hope Conference and met the Raptor reps who helped us get Kenzie on Procysbi® which has been a game changer for Kenzie's quality of life.

In the summer of 2014, Kenzie was having problems with her eyes on sunny days. In July we were sent to an opthamologist to determine if crystals were accumulating in her corneas. Crystals were found and once again we had another problem to manage. Dr. Jennifer Simpson was very helpful and started Kenzie on Cystaran™ eye drops.

WE ARE BLESSED EVERY DAY WITH KENZIE'S AMAZING SMILE AND PERSONALITY.

We thank the representatives of Sigma-Tau and Raptor Pharmaceuticals for making Cystaran™ and Procysbi® available to Kenzie. We want to thank Dr. Simpson for her help and Dr. Grimm for the information and guidance about how to treat Kenzie and a special thank you to Jeff and Nancy Stack and the Cystinosis Research Foundation.

We are blessed every day by Kenzie's amazing smile and personality. We may have a few bumps in the road but she is resilient. She never shows fear and rarely does anything bring her spirits down. She loves ballet class and playing with her big sister Brielle.

Kenzie playing doctor.

DAY *of* HOPE

By Stephen Jenkins, MD
SAM AND LARS' DAD

We just got back from another amazing Cystinosis Research Foundation family conference. There were 240 people there! With so many people and new families, the CRF actually moved locations to the Island Hotel. We missed the Balboa Bay Resort, but the Island Hotel was beautiful and very accommodating. They even gave us a discounted laundry rate to take care of our bedding every morning!

The conference started with a welcome dinner on Thursday night. We connected with old friends and met a lot of new ones. It didn't take long (about the time it takes to eat two quesadillas) before the boys were running wild with all of their friends. Sam quickly found Henry Sturgis, his favorite pal, and Lars ran along after them. Playtime was only interrupted by a few handfuls of Procysbi®, and then they were ready to keep going.

We kicked off the meeting with family introductions. Everyone stood and shared a little of their story. We all wrote down our wishes for our children and loved ones with cystinosis, and we posted them on a giant kaleidoscope heart. There were a lot of tears and laughs and hope shared. It was cool to see some new adults with cystinosis introduce themselves and share a little of their journey with the group. It felt like a big family reunion.

Nancy Stack started the next session with a talk about the Cystinosis Research Foundation, which since 2003 has raised over $33 million dollars. They have funded 134 multi-year research grants in 12 countries, with 63 publications in prestigious journals. They funded the research that led to the development of delayed release cysteamine, Procysbi®. They are the largest funder of cystinosis research in the world.

Dr. Sandra Amaral from Children's Hospital of Philadelphia attended this year and gave a talk about Fanconi Syndrome. She explained the mechanism of how cystinosis causes damage to the proximal tubule of the kidney, so it is unable to reabsorb important electrolytes, proteins and sugar. She talked about the many medications that people with Fanconi's syndrome must take, including potassium, citrate, phosphorus and others. She made the interesting point that phosphorus and calcium should not be taken at the same time because they bind each other in the gut, which impairs their absorption. Later in the conference she gave a talk about adolescents and adults with cystinosis, and the special challenges that go along with transplant, medication adherence, education, and work. She addressed strategies for coping and improving quality of life and recommended a book called "Building Resilience in Children and Teens: Giving Kids Roots and Wings" by Kenneth Ginsburg.

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Dr. Mary Leonard from Stanford University gave us an update on her study of muscle and bone health. So far, she has obtained data on 23 people with cystinosis, ages 8 to 49. Her preliminary data shows that people with cystinosis have much lower bone mineral density than average. More than half of study participants had bone mineral density less than the 10th percentile for age. She also found that people with cystinosis have significantly reduced muscle mass. More than half had less than the 5th percentile for age. She found that cystinosis bone is thinner, likely because of lack of muscle forces. She recommended weight-bearing exercise to help build stronger bone. It's also important to have enough phosphorus, calcium and vitamin D to build bone. There may also be a role for growth hormone to improve bone and muscle health. She also noted that two of the participants had unusually good bone mineral density, and this was associated with abnormal dentition. She and Dr. Grimm think this is likely secondary to fluoride toxicity. The increased bone mineral density in fluoride toxicity is actually unhealthy and is more likely to lead to fractures. Since patients with cystinosis drink such high volumes of water they may be at higher risk for excessive fluoride intake, so this is something they will look at in their study.

Dr. Mak from University of California, San Diego provided a summary on many of his studies of muscle-wasting in cystinosis. He showed his data on vitamin D. The important thing is that over-the-counter vitamin D, either cholecalciferol or ergocalciferol, also known as 25-vitamin D, may help improve muscle mass and strength. This vitamin D is different than calcitriol (1,25-vitamin D) which many people with kidney disease require for bone health.

He talked about cachexia, which is a nutritional wasting that is different than malnutrition. Even if you give patients with cystinosis adequate calories they fail to gain weight and build muscle. This process may involve the transformation of white fat to brown fat. Brown fat is something that babies need to stay warm because it burns calories to produce heat. This process is maladaptive in cystinosis because it wastes energy. Dr. Mak has shown that cystinosis mice develop more brown fat, and this is probably driven by increased cellular inflammation. His lab has found increased levels of inflammatory cytokines in cystinosis mice, including interleukin-1. They are testing an anti-inflammatory drug that blocks interleukin-1 in cystinosis mice to see whether it reduces inflammation and improves muscle mass.

Another pathway involved in cachexia is leptin signaling. Leptin is a hormone that regulates appetite and is very important in regulating energy and metabolism. Dr. Mak and his lab have treated cystinosis mice with a leptin blocker, and they found that it reversed muscle-wasting and improved muscle function. This is another exciting potential target to treat muscle-wasting in cystinosis.

After Dr. Mak, we heard from Dr. Kate Dahl, a clinical psychologist from Stanford who specializes in child and adolescent psychiatry. She talked about the ways a medical diagnosis affects every member of the family and how it can trigger distress emotions. She talked about the different ways people cope with challenges and reviewed strategies to enhance coping and communication for caregivers and people with cystinosis. She walked us through a practice run in mindfulness training, and recommended a couple of apps, including "Headspace" and "Calm." After her talk she conducted special sessions for adults with cystinosis and for caregivers of adults with cystinosis.

While Dr. Dahl did her more private sessions, the rest of us had a forum on troubleshooting many of the daily challenges of cystinosis. We talked about ways to organize medications. Some people use color coding, others lettering systems. Many families draw up enough medications for a month so syringes are ready to go anytime. Denice Flerchinger recommended Monoject Slip Tip syringes because the numbers never wear off. Nicole Manz talked about how to do a blended diet. We talked about getting a 504 plan for school in order to accommodate things like free access to the bathroom. We also talked about bedwetting, something we have continued to struggle with. I think the takeaway there was that the child will night train when they are ready, and in the mean time we should try to keep up with the laundry.

Next we heard from Dr. Bruce Barshop of the University of California, San Diego, about the new cystine measurement assay. He explained how 1.9 became our new target for cystine levels. Apparently 99.9 percent of carriers (people with one cystinosis gene) have levels less than 1.9. This number also seems to correlate very well with 1.0 and the old test. He says that his lab will still run the old white blood cell cystine test if local labs are having difficulty, but the new test should be much easier. All you need is a yellow top tube, shipped overnight to UCSD on ice. He also clarified a very important thing that was a little confusing from the original trial. Blood should be drawn 12 hours after the last dose of Procysbi® and 6 hours after the last dose of Cystagon,® and then the medication should be taken. Some patients would take the medicine and then get the blood drawn, but if there were delays in blood collection, then cystine levels could be falsely low. He also recommended that you get cystine checks at least two to three times a year, and much more frequently when converting from Cystagon® to Procysbi®.

DAY of HOPE

Betty Cabrera from University of California, San Diego talked about the importance of registering and updating our profiles on the Cure Cystinosis International Registry (CCIR). The survey has been updated with new questions that are relevant to upcoming clinical trials. It is a very important source of information for our researchers. She recommended that everyone try to update their profiles by May 1, 2016.

We capped off Friday's sessions with the adult and teen panel where we got to hear from some of the giants in the cystinosis community. We heard enlightening insights about medication compliance, moving out, working and the hope they have for a cure.

While we were at talks, the kids were having a blast with the babysitters. They








had a great itinerary, including yoga training, a magician, and a visit from some wild animals. The kids got to pet a porcupine, a hedgehog, an armadillo, an alligator, a boa constrictor, and a kinkajou! Sam loved the endless potato chips and Lars was in juice heaven.

Friday night we had another wonderful dinner, and yes, there was cotton candy with light-up wands. I think Sam looks forward to that more than anything else. He and Henry immediately set to work gathering an army of boys and declared war on the girls. There was a little bit of chaos in the hotel lobby. The whole lightsaber battle worked better on the beach at Balboa Bay . . .

Saturday morning was packed with translational research updates. We heard from Dr. Sergio Catz about a protein called LAMP2A that acts as a port of entry to the lysosome. It's an important receptor in chaperone-mediated autophagy. It's built somewhere else in the cell and has to be transported on the cellular highway to the lysosome. When the protein cystinosin is absent, LAMP2A has difficulty getting to the lysosome, and this leads to a build-up of junk outside the lysosome. This can be just as disruptive as stuff building up inside the lysosome (i.e. cystine) and leads to increased cellular stress. He is collaborating with another reseacher, Ana Maria Cuervo, at Albert Einstein College of Medicine. She has already found some molecules that stabilize LAMP2A, improve its trafficking to the lysosome and reduce cellular stress. They are testing these molecules in cystinosis mice.

Dr. Stéphanie Cherqui gave an inspiring talk about the potential for stem cell transplantation to cure cystinosis. She is almost done with the safety and toxicology studies. They have been working out the best way to transduce human stem cells with the lentivirus that holds the corrected cystinosin gene. Their protocol worked great in healthy human stem cells, but in cystinosis stem cells the lentivirus is not taken up as avidly. She is hoping to submit the IND (Investigational New Drug) paperwork to the Institutional Review Board this fall, and then we will anxiously wait for FDA approval to start the clinical trial. They will start with two adults, followed by another two adults. Then they will re-evaluate the safety of the treatment and consider two adolescents. The treatment will require a full month in the hospital, followed by weekly visits at UCSD for two-to-three months. The cure is coming!

Dr. Cherqui was followed by her PhD student Spencer Goodman. He did a fantastic job explaining the mechanism by which hematopoietic stem cells can rescue organ function in cystinosis. Stem cells turn into macrophages, which transfer healthy lysosomes to cystinosis cells through tunneling nanotubules. This mechanism holds great potential for other organelle-based diseases.

Next up we heard from Dr. Jennifer Simpson of the University of California, Irvine. She talked about how there is more to ocular cystinosis than corneal crystals. Every compartment of the eye is affected, including the retina, conjunctiva, iris and ciliary bodies. Patients with cystinosis are at high risk of dry eye because the goblet cells that secrete mucus, an important part of your tear film, are lost over time. She also noted that corneal crystals should not affect vision, so if your vision is worse than 20/30, then your ophthalmologist should look for another cause. She also talked about the risk of glaucoma, which is caused by increased pressure in the eye. This manifests as pain in the eye, redness, tearing, seeing halos, nausea and vomiting, and is an eye emergency. She also spent some time on pseudotumor cerebri, aka idiopathic intracranial hypertension, which has been seen in some patients with cystinosis. Increased intracranial pressure can damage the optic nerve, which carries visual signals from the eye to the brain. This damage can cause blindness. Any vision loss should involve evaluation of the optic nerve. She also talked about how optical coherence tomography (OCT) is superior to slit lamp exams for monitoring crystals in the cornea. She is working on cystinosis guidelines to share with our ophthalmologists.

Dr. Ghanashyam Acharya from Baylor College of Medicine updated us on the nanowafer for corneal cystinosis, which is gearing up for a clinical trial. The nanowafer is like a very thin contact lens made of polyvinyl alcohol. It is 80 microns thick, compared to a contact lens which is 200 microns thick. The nanowafer is more effective than cysteamine drops and does not need to be refrigerated because the drug is more stable. It will also improve compliance significantly. He also gave us an update on the transdermal patch, which will pump cysteamine in through the skin. It would hopefully produce more steady drug concentration in the blood and have fewer side effects. He is currently testing it on cadaver skin and pigs!

The final speaker was Dr. Doris Trauner from University of California, San Diego, who summarized her findings of her study on quality of life and psychosocial functioning in teens and adults with cystinosis. She found that adults and teens with cystinosis have problems with sleep, anxiety, depression, fatigue and independence. They also reported strong emotional and family support.

We concluded the session with a Q&A panel with the physicians and researchers. As in previous years, people expressed interest in doing research on male fertility. There were several questions about medication compatibility. Procysbi® should be taken with acids, like orange juice, and should not be taken with bicarbonate.

Saturday night was the big Natalie's Wish event. Twenty-one families presented checks to the CRF this year! The CRF brought in a record $3.3 million dollars that night, and the money keeps coming in!

This year, Rachel Platten, popular singer of "Fight Song" provided the entertainment. She met the kids before the event and took pictures. At the end of the gala she had all the kids come up to the stage to sing "Fight Song." There were a lot of tears. It was the perfect end to the perfect conference. We all left energized to keep fighting cystinosis.

DAY of HOPE

A Community United By Hope

By Polly Knelsen, family friend
LEWISTON, IDAHO

The consequences of a very rare, complicated and (at this time) incurable disease go much further than the afflicted child. Parents, siblings, grandparents, school friends, and, in our case, the entire community have been affected.

Tina Flerchinger and her family have been dealing with a cystinosis diagnosis since 2005, but it wasn't until 2008 that I heard about Tina. I knew the Flerchinger family a little in that we always greeted each other at community events and shared smiles and laughs at our mutual friends' weddings and parties. I knew they were a good family, strong in their faith, with three gorgeous daughters. I knew we had common hopes and similar philosophies. What I didn't know until attending their fundraiser in 2008 was the kind of strength and endurance this precious family has displayed during the fight for their daughter's life. It's hard to believe that it was eight years ago, as I now hold this family deep in my heart and daily in my prayers.

In May of 2015 our community took part in the seventh annual Wine, Stein, and Dine event benefitting "Tina's Hope for a Cure." The tagline on the invitations read, "please join family and friends for an evening of hope." Admittedly, by the day of the event, it may have been more appropriate to say "please join Tina's family and friends for a long nap, they are utterly exhausted!" as they all were weary after planning such an event. The show was a great success. Over 250 people attended and showed support, not only financially, but in the hugs and kind words expressed to Tina and her family throughout the evening. The apex of the evening is also the toughest moment of the evening. This is when we learn more about the specifics of what Tina and her family face every minute of every day. It's hard for many to grasp what it is like to take roughly 50 pills a day just to stay alive.

This is Tina's life. This is her family's life. This is hard. I calculate that Tina's parents, Denice and Mark, have walked this road of care and compassion – together with a plethora of meds – with their youngest daughter for more than 4,000 days…

The Flerchinger family has one goal: Find a cure for Tina. The road to a cure is not a straight line. They approach every day with cautious optimism. I think the word "optimism" is somewhat perfect to describe this family, especially Tina. Every year brings her to a new level of maturity and a better understanding of her disease. But, she does not waver in her zest for life. She wants to do just what all other kids do. She wants to get up and go to school and to have a "normal" life where possible. Tina and her family have taught our community much about living with optimism. Because of their quest for a miracle, they encourage us all to continue to think big and invest time and resources in ambitious plans. They live their lives in a way that makes our community want to keep giving and helping the Flerchinger family reach their goal to find a cure for Tina.

THE ROAD TO A CURE IS NOT A STRAIGHT LINE. THEY APPROACH EVERY DAY WITH CAUTIOUS OPTIMISM.

Polly Knelson and Tina

The Village who helps our family every day!

By Brian and Tricia Sturgis, Henry's parents
SANDPOINT, IDAHO

THE FOLLOWING PEOPLE HELP US ON A DAILY BASIS AND WE ARE SO GRATEFUL FOR THEIR FRIENDSHIPS.

FAMILY: Our families are a huge support system for us. Henry has two grandmothers and one grandfather that live in Sandpoint, Idaho, the same town we live in. They help with picking up Henry after school and whatever else needs to be done so that we can both work a full day. Brian's sister and brother-in-law also live in Sandpoint and are always willing to lend a hand. We are so lucky to have our family nearby to help take care of Henry, it's a blessing. They know what his needs are and they have the patience and love to help him through every day.

FRIENDS: We are so grateful for friends, old and new! It is really uplifting to know that our friends care about Henry, our family and finding a cure for cystinosis. Some days it's hard to put your best face forward, and when we can't, our friends are often the ones who keep us going, keep us stable and grounded and motivate us to be our best.

24 HOURS FOR HANK: We love the foundation we have created over the last eight years and TOGETHER we have made it a success. Our fearless leader and board president, Rob Timmons, leads our board with drive and determination. He knows Henry's day-to-day struggles and help's rally the troops to raise money to find a cure for cystinosis. He also raises our spirits and keeps us going with his humor and big heart. We would also like to thank ALL of our board members and their families for their countless hours spent working on 24 Hours for Hank.

CRF: Board members, researchers and scientists — the Cystinosis Research Foundation is a huge source of our day-to-day well-being by offering hope, community, education and access to the best doctors in the world. They work tirelessly to help all of the families with many issues we face. We love being able to make great connections with cystinosis families that we see every year, many have become great friends. Henry would not have received such great care, since the beginning of his diagnosis, without the brilliant researchers and doctors who bring their science and studies to the families and medical communities within their expertise. These further developments in medicine affect more diseases than just cystinosis, many breakthroughs can be transferred to other diseases.

THE STACK FAMILY: I think of Natalie Stack's wish when she wrote it on a napkin when she was 12. Her courage and bravery in helping others affected by cystinosis is a true gift. She inspires us, just as her parents Nancy and Jeff do. It takes perseverance and resilience to keep fighting this fight- they haven't given up and neither will we- we can't thank each of them enough!

HENRY'S DOCTORS AND THERAPISTS: We couldn't do it without Henry's doctors, nurses, therapists, pharmacists, lab techs and hospital staff that help us facilitate special tests that Henry needs to have done and meds that need to be compounded. It takes a village and we thank you ALL for your sacrifices and support, we couldn't do it without you!

CYSTINOSIS COMMUNITY

A Letter To My Former Self

By Erin Little, Olivia's mom
PORT ELGIN, ONTARIO, CANADA

The following is a letter to my former self, with all the things I have learned since 2011 when Olivia was first diagnosed.

Dear Former Self,

Dr. Filler just shared some devastating news. You haven't even Googled it yet - probably because you can't even spell it, let alone pronounce it. Little do you know you'll soon be explaining the ins and outs of this disease, and the spelling of it, so that it's not misprinted in your local newspaper.

First and foremost, I want to tell you this is not your fault. You did everything in your power to find answers to your daughter's ongoing health issues. You attended countless visits to your family doctor sharing your concerns about her water intake, poor growth, slow weight gain and lack of appetite. You have been in and out of the emergency room three times within a week and still no one listened. You followed your gut and visited the emergency room one more time.

You saved your daughter's life. You will think about how devastating it could have been if you didn't push yourself to bring her in that one last time.

You will be in the hospital for a month and will meet a lot of amazing people who are going to be a huge part of your family's life. Listen and learn from them, they are going to become like family to you. Do not let your differences blind you from your similarities.

Coming home is going to be difficult and your support will be limited. Don't focus on those who won't be there for you; embrace those who are. From this day forward it will be about the three of you. Accepting help is going to be hard, but make sure you surround yourself with people who are willing to be there for you. Welcome what they will have to offer.

Your choice to focus on your family and what Olivia needs will be tough, but it will allow you to organize your priorities and adjust to your new way of living. The mother that you first reach out to that focuses on all of the negatives will be a blessing in disguise. Never forget what it feels like to be on that end of that phone call, with a newly diagnosed kid. Stay positive and focus on the now, not what may or may not happen. When that phone conversation doesn't go the way you anticipated it, it's only because the timing is wrong. You will be heartbroken, but please know that very soon you are going to find an amazing organization that will change your life. It's called the Cystinosis Research Foundation. The CRF is going to bring incredible new hope to you and your family.

The constant vomiting will subside, and you will master how to deal with those issues. You will have moments that will break you, but in the end they will make you stronger. You will find an answer to one problem and another will emerge. But you will solve it. If you can't solve it alone, you will reach out to your beautiful cystinosis community.

One day you will be grateful for this awful diagnosis. In the end, you can never change how all this happened, but you can choose how you view it. Embrace it. Your "gratitude attitude" for all the things cystinosis has brought into your life will be difficult to swallow, but it holds truth. You will meet new friends who become some of your best friends, and you will have more in common with them than just cystinosis. Your marriage will become stronger than you ever thought it could be, as you will live the vows that you once stood and said in front of family and friends. You will have a new appreciation for what family means, and you will learn that the small things are what matter most - the smiles, the laughs, and all the cuddles in between. You will find out how quickly they can be taken away from you.

> ***LIVE IN THE MOMENT.***
> ***STOP AND SMELL THE ROSES.***
> ***FOLLOW YOUR DREAMS.***
> ***ALWAYS REMEMBER TO LOVE.***

Always believe and never let go of hope. Hope will keep your family together, and when things get tough – remember, anything is possible. If you can imagine it, it can happen. You have the best team in the world fighting for cystinosis.

And always know, you are an incredible mother. Olivia chose you and Chad to guide her; to do what's best. She will thrive. She will do amazing things with the life she is given. In the end, you will always be moving forward, with the best intentions, doing what you know is best.

Like when you followed your gut and visited the ER that one last time. You will save your daughter's life.

Love and big hugs,
Erin Little (today)

I CAN'T EVEN TELL YOU HOW AMAZING IT IS TO WORK ON CYSTINOSIS PROJECTS, HOPING THAT I CAN MAKE A DIFFERENCE FOR MY SISTER AND FOR EVERY OTHER CHILD AND ADULT LIVING WITH THIS DISEASE.

From Bedside to Bench to Bedside

By Tatiana Lobry, Wendy's sister
DEVILLE, CHAMPAGNE-ARDENNE, FRANCE

When I was 6 years old, my baby sister Wendy was born. Early on she stopped eating and started drinking a lot of water. Because my sister was the fourth child, my parents had gained enough experience parenting to know something was different with her. This began the year of doctor visits, hospital stays and lab tests. That was how I first learned the word cystinosis.

My mom and dad had already lost a son to sudden infant death syndrome (SIDS), so this must have been a new devastating blow to them as parents. The daily routine at home now consisted of pills, growth hormone injections, esophageal feeding tubes, more doctor visits and watching my sister go through many bouts of nausea. Six years later, Vaincre les Maladies, an organization that planned vacations for families with children who had lysosomal disorders, sent us on our first-ever family vacation. There we met other families with children who had rare diseases. We also met a researcher by the name of Dr. Corinne Antignac, who was working on cystinosis in Paris.

I am very proud of my little sister because she never gives up. I know she has gone through hard times with the nausea, pill regimen, shots, doctor visits and even at school from one of the most noticeable side effects of cysteamine: the odor. She went through many surgeries to fix her genu valgum and, most recently, my mom was able to give her a kidney. During this time, I was far away at college and couldn't be at my sister's bedside. What a relief when the phone rang and I could hear my mom, dad and sister's voice letting me know the surgeries went well. It is now two years and six months later, Wendy is almost 20 and she is still doing great. She has graduated high school and is attending the local college.

It was because of this experience that I chose to study genetics in college. It has always amazed me that something as small as a gene can decide the color of your eyes, the color of your hair and if you have cystinosis or not. During my studies, I found an article written by Dr. Stéphanie Cherqui in 2009 titled "Successful Treatment of the Murine Model of Cystinosis Using Bone Marrow Cell Transplantation." After reading this article, I remember thinking, "Could this be the end of cystinosis?" A few months later, my family and I attended the seventh International Cystinosis Congress in Paris where I was able to meet Dr. Cherqui. Fast forward a few more years and now I am a graduate student in Dr. Cherqui's lab at the University of California, San Diego (with a fellowship from Vaincre Les Maladies Lysosomales). I can't even tell you how amazing it is to work on cystinosis projects, hoping that I can make a difference for my sister and for every other child and adult living with this disease.

My family was excited when I told them that the Cystinosis Research Foundation asked me to write an article about our story. I asked Wendy if she wanted to say something and I will end this article with her words: "It feels good to know that we are not alone and that all over the world there are people fighting to find a cure for cystinosis. Thank you, Cystinosis Research Foundation, and to all the researchers, for what you are doing."

BEING HONEST WITH OURSELVES AND REMAINING

Hopeful, Optimistic & Happy

SOLD-OUT EVENT!

SWING GOLF TOURNAMENT
Thursday, March 10th, 2016
Catta Verdera Country Club
Lincoln, California

BLING AUCTION & DINNER
Friday, March 11th, 2016
The Citizen Hotel
Sacramento, California

By Kevin Partington,
Jenna and Patrick's dad
SACRAMENTO, CALIFORNIA

On March 30, 2006, our children Jenna and Patrick were diagnosed with cystinosis. After meeting with doctors, spending countless hours researching the rare disease and speaking with other families who have been afflicted with cystinosis, we decided to form a foundation in partnership with the Cystinosis Research Foundation called Jenna & Patrick's Foundation of Hope, Inc.

Each year our family hosts Swing & Bling, a golf tournament and gala to support cystinosis research. And each year I trick myself into thinking that it is going to be easier to deliver a meaningful speech sharing our family's journey with cystinosis. It never does. In fact, it only gets more difficult as our relationship with our children and other families in the cystinosis community grow. We are truly in a race against time.

My wife, Teresa, and I would like to express with honest and realistic detail a snapshot of what life for our family is truly like. Our emotions run the gamut: from gratitude for the people we have met, to intense fear of the disease that affects Jenna and Patrick, to cautious hope for the research being funded and accomplished. I approach my honesty with caution, because this is not the side of me typically seen by friends and family who know my love of life and determination to remain ultimately hopeful, optimistic and happy.

THE TRUTH IS ...

1 The truth is…we have lost five people with cystinosis between the ages of 21 and 28 in the past several months. This is a high number considering there are fewer than 600 patients with cystinosis in the United States.

2 The truth is…on the outside our children look generally healthy, but on the inside, cystinosis is destroying every organ in their body in spite of the myriad medications they take. In the past year Jenna and Patrick have visited medical specialists in nephrology, ophthalmology, orthopedics and psychology, due to the side effects of cystine crystal accumulation.

3 The truth is…cystinosis has shown us just how loving family, friends and strangers can be. Strangely, our burden has brought our family closer to a great number of people both inside and outside the cystinosis community. We are grateful for these relationships and recognize this silver lining.

4 The truth is…our donors have helped fund every bench and clinical study that led to the discovery of a delayed release medication, approved by the FDA in 2013. Our donors have helped fund the first donor stem cell transplantation trial, currently open at UCLA. There is now a nanowafer eye treatment a year from clinical trial. The same scientist who developed this nanotechnology is also working on a transdermal patch, which could change the way cystinosis drugs are delivered, thus eliminating many side effects and doses of pills throughout the day. This research, funded by our donors, gives us hope.

5 The truth is…Jenna and Patrick both take roughly 45 pills each day; or 315 pills a week, or 16,380 pills a year. They take eye drops three times a day. That is nine doses short of the every waking hour recommendation, but its all we can deal with right now.

6 The truth is…we worry about our kids socially as they enter their middle school and high school years. We feel the "age of innocence" coming to a close. The medicine they take to extend their lives has a sulfuric smell that comes from their pores and their breath and it can overwhelm those who are near them. The 4 or 5 gallons of water per day that Jenna and Patrick require continue to be a challenge. Jenna, in particular, is so often tired and thirsty, making each day unpredictable. As we look toward the future we consider the fact that a male cystinosis patient has never fathered a child. Naturally, this is a facet of this multi-systemic disease that we are eager to see researched.

7 The truth is…Our community is our support, our village, our people! The parents of Jenna and Patrick's friends and their fantastic teachers have done an amazing job helping other children and students understand what makes Jenna and Patrick unique. Our kids feel happy and loved at school, and it is in no small part because of their compassionate and understanding friends and teachers. They are each a part of our team, giving comfort and showing understanding, and ultimately loving our children as their own family.

8 The truth is…Teresa and I are not the only ones with heartache. We are all fragile, and each of us needs the love and support of others. It is important to us that our plea for support does not take away from the plight of people we love who are dealing with cancer, epilepsy, multiple sclerosis, autism, autoimmune disorders, depression and a myriad other health challenges. The time we share with them fills our hearts and holds us over during the inevitable days when we're not so put together, when we may not be so well fed, and when our hearts might not be so full.

9 The truth is…We've come to the conclusion that it isn't supposed to be easy. We are called upon to have the courage to find support in the most meaningful, healthy ways we can. As we tell our story tonight, we realize how blessed we are to be able to be so honest, and to have this forum to share what is difficult, and to receive such support for it. We are so thankful for the support we have received as we try to make a difference in the lives of our children and many others who will be touched by the medical progress that gives us hope each day.

SAVE THE DATE

Natalie's Wish Ninth Annual Fore a Cure Golf Tournament

Monday, October 17, 2016 *at the world-class Pelican Hill Golf Course in Newport Beach*

Since 2007, the exceptional support and generosity of our friends and the community has contributed more than $2.6 million to cystinosis research which has enabled CRF to make significant advances in the treatment of cystinosis. Our dedicated committee and volunteers are working diligently to ensure a memorable golf tournament experience for Orange County's top business and community leaders.

In 2015, our Fore a Cure tournament was ranked as one of the Top 5 Golf Tournaments for the third year in a row by the Orange County Business Journal. Make your plans now to join us for a magnificent day of golf, camaraderie, and an opportunity to make a wish come true for those affected by cystinosis.

THE RESORT AT PELICAN HILL NEWPORT BEACH

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

SIBLING LOVE:

The Greatest Ally of All

By Karen McCullagh and Don Cunningham, Andrew's parents

LANGDON, ALBERTA, CANADA

Our daughter Kelsey was only 13 years old when she looked deep into my eyes and said "Mom. Breathe you are almost there." I was in labor with her little brother Andrew and she was a steady, patient and compassionate coach. She saw Andrew make his way into the world and fell in love with him from the minute she held him in her arms. Fifteen months later, he was diagnosed with cystinosis and she made a secret promise to always be there for him. She pulled his meds, cleaned his vomit, changed his diapers and his bed, fed him, bathed him, loved him and vowed to always be there for him.

She also swore she would give him her kidney should it ever come to it and spent her adolescent years avoiding anything she believed might impact her brother's kidney. "As far as I'm concerned, I only have one kidney, the other one is Andrew's so I better take care of it," is what she would tell anyone who would listen. She raised funds for Kidney March, an annual 100-kilometer walk over three days, and continually promoted the importance of organ donation. She signed her donor card at 18 and has always made her intentions known.

In December 2015, Kelsey moved to Ireland to follow her own dreams, but her brother was never far from her thoughts. She applied to become a volunteer for Cystinosis Ireland but her passion and knowledge convinced them that she was capable of so much more. In March, she was hired to work for Cystinosis Ireland and turned her education and passion into a job. She has always been Andrew's greatest ally, which prompted us to start a year-long campaign called "$11 for 11 – Allies for Andrew," which celebrates 11 years of post-diagnosis for Andrew. We have reached out and asked supporters to donate $11 dollars in Andrew's honor to support cystinosis research. We have also linked the online campaign into our 5th Annual JCFG Charity Golf Tournament, which will take place on September 10th, 2016 at Boulder Creek Golf Course in Langdon, Alberta Canada.

Throughout this journey, we have been blessed. We are surrounded by the love and support of families around the world. Many strangers have turned into our friends, who support our efforts to raise funds. We have seen the generosity of the human spirit as our circle of allies has grown from year to year, but most importantly, we bear witness to the power of sibling love. We are so blessed to have an amazing daughter like Kelsey, who has so much love and compassion for our wonderful son Andrew.

CYSTINOSIS COMMUNITY

BRIAN STURGIS

Brian Sturgis is currently the director of business operations at SimulStat Incorporated, a San Diego-based corporation that provides bio statistical programming services to pharmaceutical and biotechnology industries. Before starting at SimulStat, Brian was the director of online marketing for women's retailer, Coldwater Creek. Brian earned his degree in business and marketing from Shoreline College in Washington. Brian and his family currently reside in Sandpoint, Idaho, a small resort town in Northern Idaho, and enjoy spending time outside skiing, camping, biking and on the lake.

Brian's connection to the cystinosis community began in November 2007, when his son Henry was diagnosed with cystinosis at 16 months old. As an infant, Henry was in the 90th percentile. However by 16-months old, he was no longer on the charts and showed signs of failure to thrive. After insisting on additional tests, Henry's pediatrician sent lab results to the University of Washington where they were analyzed by Dr. Hahn. Dr. Hahn's mentor, Dr. Gahl, had done extensive research on cystinosis. When Henry was diagnosed, he began a regime of six medications and a six-hour dosing schedule of Cystagon®. Soon, Henry began to improve and started to bear weight and walk. The Sturgis family was blessed to be connected with a cystinosis family, the Flerchingers, within weeks of Henry's diagnosis. The Flerchingers live three hours away and offer the Sturgis family overwhelming support and understanding.

Tricia, Brian and Henry Sturgis

Brian looks forward to serving as a trustee on the board for the Cystinosis Research Foundation. Brian hopes his experience in business as well as raising a child with cystinosis will positively impact the Cystinosis Research Foundation and provide hope for new families coping with the diagnosis of cystinosis. Brian is passionate about finding better treatments and ultimately a cure for cystinosis.

BOB OLSON

Robert D. Olson is the founder of R.D. Olson Development, an Irvine, California-based firm engaged in the development and repositioning of commercial properties nationwide. He established the company in 1997 following nearly 20 years as founder and CEO of R.D. Olson Construction.

Robert guides the strategic vision for R.D. Olson Development with an active, hands-on approach toward every facet of construction and development. He works closely with design and development teams in some of the company's most prestigious projects, providing valuable input for marketing, investment analysis, debt and equity structures, planning and entitlements, budgeting, constructability and end-user requirements.

Robert Olson has been recognized as a business leader and entrepreneur on numerous occasions. He is the 2016 Robert Mondavi Wine & Food Award winner. He has also been the recipient of Ernst & Young's Entrepreneur of the Year award, Construction Financial Management Association's Construction Executive of the Year and the Orange County Business Journal's Excellence in Entrepreneurship award. He was recognized as a 2012 Hotel Icon by Real Estate FORUM magazine, and is a frequent speaker at leading industry events.

Robert is an active member of the Young Presidents' Organization (YPO), including having served as Chairman of the California Coast Chapter. His philanthropic activity includes serving on the Board of Directors and Executive Committee of the Orange County Chapter of the American Red Cross. Robert is an MBA graduate of the University of Southern California.

Charlotte, Sutton, Bob and Christyne Olson

Long-time friends of Nancy and Jeff Stack, Robert has admired the Stack family's dedication to finding a cure for cystinosis. He is inspired by families that are affected by the disease and wants to move mountains in any way possible. Robert hopes his leadership skills and business understanding will help guide the organization through the future and provide hope for the many families afflicted with this disease.

DAVID MOSSMAN

"I am humbled at the opportunity to join in the quest for the cure!"

David W. Mossman currently serves as the executive vice president and chief investment officer for Donahue Schriber Realty Group, L.P. In his 20 years with Donahue Schriber, David has overseen in excess of $2 billion of new property acquisitions, property dispositions, and development projects for the company. Prior to joining Donahue Schriber, David served as a manager for Deloitte.

David is a Certified Public Accountant (CPA) and graduate of California Polytechnic State University, San Luis Obispo. He is also an active member of the International Council of Shopping Centers (ICSC). David believes in volunteering his time to organizations which help children prosper both academically and as individuals. He has served as a board member for Harbor Day School in Corona Del Mar and KidWorks in Santa Ana from 2012-2015, and was a trustee for The Pegasus School in Huntington Beach from 2005-2011.

David and his wife, Bianca, became connected with the Cystinosis Research Foundation when their friends, Kevin and Teresa Partingon's twins were diagnosed with cystinosis. David and Bianca are passionate about supporting those affected with cystinosis. He is inspired by what has been accomplished to date by the Cystinosis Research Foundation and looks forward to contributing to the organization's continued progress.

Sophia, Bianca, Caden, Drake, Colton and David Mossman

MARCU ALEXANDER

Marcu Alexander is a dedicated wife and mother of two young daughters. Marcu resides with her family in Boise, Idaho. She is a graduate from Western Washington University with a bachelor's degree in psychology and a minor in sociology. Marcu currently holds the position of packaging development manager for Happy Family Brands. Happy Family Brands is a mom-founded and operated organic food company that has changed the way families feed their children. Marcu advocates for wellness at Happy Family Brands and speaks on behalf of the company at various community events.

Marcu's youngest daughter, Hadley, was diagnosed with cystinosis in April of 2012 at 18 months old. Since Hadley's diagnosis, Marcu and her family have prioritized involvement with the cystinosis community and stay updated on the latest technology and research. The Cystinosis Research Foundation has introduced the family to other families going through the same experience, who have made this journey far less scary and isolating.

Marcu is the founder and president of the non-profit group, Hearts for Hadley. Hearts for Hadley is dedicated to spreading awareness and raising research funds for cystinosis. Hearts for Hadley donates 100 percent of its proceeds to the Cystinosis Research Foundation. As the leader of Hearts for Hadley, Marcu organizes an annual Hearts for Hadley fundraiser which has received great support from the Boise community. In addition, Marcu has helped organize additional fundraising events including Kegs4Kause and a New Year's Eve event.

Marcu and her family attended their first Day of Hope family conference two weeks after Hadley's diagnosis. Since then, the family has been present at all subsequent Day of Hope conferences and is committed to attending every year. Marcu also engages in cystinosis support groups and is a regular contributor to the Cystinosis Magazine.

Marcu is driven to continue learning more about cystinosis research and advocating for the community. As a member of the board of trustees, she will bring positive energy, passionate enthusiasm and strong commitment to the other board members and leaders. Marcu's greatest ambition is to spread awareness and work tirelessly to raise funds for research that will provide hope and a cure to those living with this disease.

Marcu, Hadley, Ben and Stella Alexander

YOUR VOICE MATTERS

You Can Help Find the Cure!

Visit the CCIR website today:

WWW.CYSTINOSISREGISTRY.ORG

CURE CYSTINOSIS INTERNATIONAL REGISTRY

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 519 registrants from 43 different countries. 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.

To date, 51 new surveys have been completed, representing new and established registrants alike. If you have not yet had the opportunity to take the expanded survey, please take 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.

CURE CYSTINOSIS INTERNATIONAL REGISTRY

Intended to capture richer, more detailed information about cystinosis … necessary for advancing therapies.

ABOUT THE EXPANDED CCIR SURVEY

The survey now includes additional questions, several of which ask about an affected person's experience with the various cystinosis treatments now on the market, such as Procysbi® and Cystaran™®. A subset of existing survey questions has been amended so that there are more answer options to choose from.

If you have previously completed the older version of the CCIR survey, you will notice that the answers you provided to existing or unmodified questions are still recorded in the system. Please check that the answer options you marked are still accurate today. If not, please update your response.

New or modified questions will be obvious as there will be no answers marked. Please provide answers to these questions.

If at any point, you encounter any difficulties and require assistance, please contact the registry Curator at *curator@cystinosisregistry.org*.

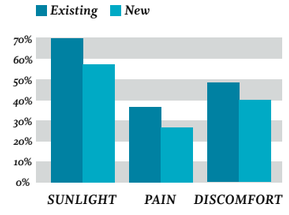
EXAMPLES OF NEW SURVEY QUESTIONS

- *Has the affected person experienced difficulties with speech due to weakened vocal muscles?*
- *Have speech difficulties significantly impacted the affected person's ability to communicate with others?*
- *Is the affected person followed by an eye care specialist?*
- *If affected person is currently receiving care by one or more pediatric specialists, have you spoken with them about transition to adult care?*

OPHTHALMOLOGIC

50 out of 65 patients report receiving eye care, and half have annual visits. The most prevalent ophthalmologic symptoms are trouble seeing in sunlight, pain and discomfort. The new survey elicits 13 additional symptoms, with the most prevalent of these being headache (12), burning sensation (9) and sandy or gritty sensation (8). Cystaran™ use is captured and is considered helpful overall.

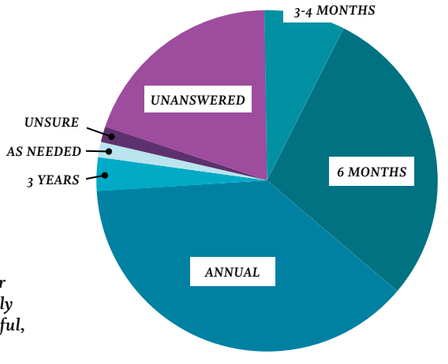
MOST PREVALENT SYMPTOMS



CYSTARAN™® EYE DROP USE (n=65)

Overall usage (n=65): 28 patients ever used Cystaran™ drops, and 23 currently use them. 19 reported drops to be helpful, and 1 reported drops as not helpful.

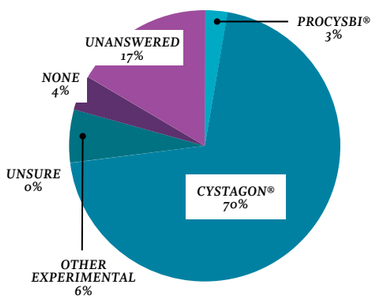
FREQUENCY OF EYE CARE VISITS (n=65)



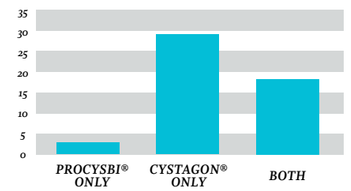
ORAL TREATMENT

Overall, most patients are currently treated with Cystagon® In the subset of patients who completed the revised survey and live in countries where Procysbi® is available, approximately two-thirds have been treated exclusively with Cystagon® and one-third with both Cystagon® and Procysbi® in their lifetime.

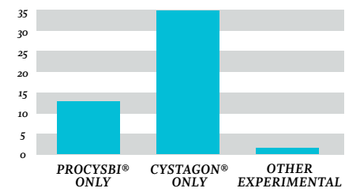
CURRENT TREATMENT (n=512)



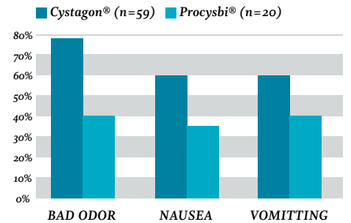
ORAL TREATMENT(S) EVER RECEIVED (n=49)



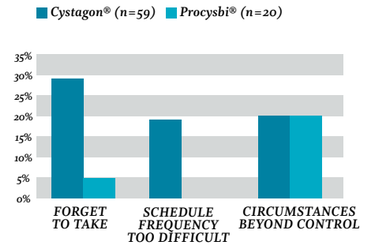
CURRENT TREATMENT (n=49)



MOST FREQUENT REPORTED SIDE EFFECTS



MOST FREQUENT REPORTED REASONS FOR NON ADHERENCE



Out of 65 patients, only 49 live where Procysbi® is approved.

TOGETHER, WE ARE one

1 PURPOSE.
1 JOURNEY.
1 CURE.

BAILEY DEDIO

BAILEY FEST

Bailey Believes
HIS HOPES, HIS DREAMS AND A CURE

The annual Bailey Believes Fest, held in honor of Bailey DeDio, was hosted this year at the Lucky John's Bar & Grill in conjunction with Doggie Stylin' and Grooming Salon in Stanton, California. Jan Webb and Josanna Charity, owner of the grooming salon is a family friend who helped rally the local community to come together and donate items for the silent auction and participate in the day of fun, food, drinks, and music in honor of Bailey. Together they raised more than $5,785 to help find the cure for cystinosis.

HADLEY ALEXANDER

HEARTS FOR HADLEY

The Hearts for Hadley New Year's Eve Celebration welcomed in 2016 with fun, food and the legendary garage band Doon! The festive evening, held in honor of Hadley Alexander, included heavy appetizers, beer and wine and raised more than $3,150 which will help fund research to make a difference in the lives of the children and adults with cystinosis. Thank you to the Alexander Family and the Boise, Idaho, community for their support in our quest for the cure.

LANDON HARTZ

The Hartz family hosted the 5th Annual Lots of Love for Landon Halloween Fundraiser at the Castle Shannon Fire Hall in Pittsburgh, Pennsylvania which once again was a very successful event. The 350 festive party-goers enjoyed tricks and treats, great music, a costume contest, Chinese auction and special door prizes. Held in honor of Landon Hartz, the participants contributed more than $12,300 to support cystinosis research for improved cystinosis treatments and a cure!

Lots of Love for Landon
A HALLOWEEN GATHERING

ANDREW CUNNINGHAM

J.C.F.G. MEMORIAL GOLF TOURNAMENT

The 4th Annual J.C.F.G. Memorial Golf Tournament held in Alberta, Canada, was formed in memory of 'Fore Fathers' from the McCullagh, Cameron, Halluk and Cunningham Families. The event held in honor of Andrew Cunningham, raised more than $19,592 for cystinosis research. An additional $10,525 was collected from the event website and will be added to the funds raised from previous events with other Canadian families in the Aqueduct Foundation's Canadian Cystinosis Research Fund which works in partnership with CRF to fund research.

BROOKE EMERSON

FISHING FOR BROOKE'S CURE

While Brooke was only diagnosed a few short months ago, Clay and Jill (Brooke's parents) felt that a fundraiser would help them cope with the diagnosis while at the same time allowing them to raise funds for cystinosis research. Unlike other fundraisers they'd supported, they couldn't survive a 5K and they hadn't ridden a bicycle since the 7th grade. Clay has fished his whole life and fishing is just one common element that he and his friends have in common, so he roped in three of his friends and the first annual Fishing For Brooke's Cure was born!

On Earth Day, April 22, 2016, Clay and his friends set out to some local rivers in Delaware and Maryland to fish for shad, a migratory fish that lives in the ocean and can be found in freshwater rivers for a brief period of time in the spring. The rules were simple; each of the four anglers spread the word by email, social media and word of mouth, and together were able to round up nearly 100 sponsors, each of who pledged a certain amount per fish caught with an optional "not to exceed" amount.

The four anglers fished from sunrise to sunset, and the fishing was better than imagined, with all four anglers having their best day on the water and a total of 335 fish caught (and released). To everyone's delight, the event generated over $16,000 in pledged donations!

Clay, Jill and Brooke Emerson are so happy to partner with the Cystinosis Research Foundation and donate these funds to finding a cure and better treatment for Brooke and the other children and young adults in the cystinosis community.

JENNA AND PATRICK PARTINGTON

BUNCO NIGHT

Jenna & Patrick's FOUNDATION OF HOPE

A Bunco Night to Benefit Jenna & Patrick's Foundation of Hope was recently held at Husick's Taphouse in Clarksburg, California, and was organized by Saralyn Taormina, whose son Leo is a 5th grade classmate of Jenna and Patrick Partington at Holy Spirit Parish School.

Saralyn's monthly Bunco group selects a different local charity each month and Jenna & Patrick's Foundation of Hope was her choice for the April gathering. Participants at the event were moms from Holy Spirit, friends from Cushman and Wakefield, and community friends who came together to support JPFH. The evening provided a wonderful opportunity for people to network and get to know each other better and learn more about cystinosis, the disease affecting Jenna and Patrick. At the end of the night more than $800 was raised to help fund research.

SHANNON KEIZER

SHANNON KEIZER'S BIRTHDAY SOIREE

April 23, 2016 will go down as one of the most memorable nights of my life so far. With the help of many friends and family, I hosted my first cystinosis fundraising event. And most importantly, stories were shared and lives were touched.

I've always admired those who put on big events each year. I often thought of doing one myself, but didn't think I had what it takes or that anyone outside my family and close friends would really care what I had to say. That all changed when I moved to Seattle about seven months ago.

Since moving, I've gained self-confidence and discovered my voice. While attending a professional development training with about 150 other people, I stood up and shared my life story to a room full of strangers. There was not a dry eye in the whole place. People were moved and inspired. In that moment I realized I had a gift and a voice, and I needed to do something with it.

I talked over the idea of a birthday fundraiser with my dad. He immediately called his sister, my Aunt Mary, and asked her to fly out from Michigan to make a salad (her salads are quite bomb for large groups of people). What began with a salad turned into a whole menu, which turned into desserts, table decorations, and task delegating. With one phone call I suddenly had a team of people that were committed to making my dream a reality.

This process allowed me to become close with the Head family, who live about an hour and a half south of my Seattle home. Melissa and I met at a coffee shop and discussed the process of hosting a fundraiser. An immediate friendship was formed. After spending Easter with the Heads, their kids began referring to me as another aunt, and I had a second family.

After months of preparations, my Michigan family (parents, aunts and uncles) flew in, and the days of chaos began. That famous phrase, "It takes a village," couldn't be more true. I am so thankful for such wonderful friends and family who work together to accomplish something bigger than any one person.

The night was a success. It began with "Seattle's Baritone Vocalist," Ken Sabalza, singing inspirational songs from Frank Sinatra to "How Great Thou Art." My dad, Kevin Keizer, emceed the night and shared the powerful story of my diagnosis. Melissa Head talked about the CRF and her daughter Mary's story. I completed the program speaking about my life and the lessons I've learned in my 27 years living with cystinosis. Lives were touched, and people were moved. I wish I could put into words the magic that happened that night, but it would take at least another three pages to just touch the surface. The evening completed with a performance of "You Raise Me Up" and audience participation in "My Girl." People left wanting more and wanted to know what's next. They were on fire. Days later, I'm still hearing about the impact it made in so many lives. At this point, $8,300 has been raised and funds are still coming in!

I would like to thank Nancy, Zoe, Stacy and the Cystinosis Research Foundation for making all of this possible. Thank you for giving so many people a voice and all you do towards the development of cystinosis research. And thank you to my friends and family who have come along side the CRF in our cause.

My dream is to be a motivational speaker and allow my story to transform lives around the world. That dream began this past weekend. I've already began planning next year's event, which will be held in Michigan. So Michigan friends and family, be ready! I look forward to continuing this journey with a community of such wonderful people at my side.

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.

CHAIR

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

BOARD MEMBERS

Daniel G. Bichet, MD, MSc
Professor of Secondary Affiliation (Medicine)
Department of Molecular and Integrative Physiology
Faculty of Medicine, Université de Montréal
Centre de recherché, Hôpital du Sacré-Coeur de Montréal
QUEBEC, CANADA

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de Duve Institute
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Center for Clinical and Translational Research
Professor of Pediatrics
Ohio State University
COLUMBUS, OHIO

Thank you for your dedication to the global cystinosis community.

CYSTINOSIS RESEARCH FOUNDATION WELCOMES TWO NEW SCIENTIFIC REVIEW BOARD MEMBERS

Daniel G. Bichet is Professor of Medicine and Molecular and Integrative Physiology at University of Montreal and a nephrologist at the hospital of Sacré-Coeur in Montreal. He obtained his medical degree at the University of Besançon (France) and completed additional clinical training at University of Montreal and McGill University affiliated hospitals. He did a research fellowship at the University of Colorado Health Sciences Center under the mentorship of Dr. Robert W. Schrier.

He received the Jonathan Ballon Award of the Quebec Heart and Stroke Foundation and obtained a Canadian Institutes of Health Research Chair in Genetics of Renal Diseases from 2003 to 2010.

Dr. Bichet's research includes fundamental life-sustaining homeostatic networks for water and osmotic pressure balance in human physiology. His laboratory is contributing to the prevention of extreme dehydration states in children with polyuric disorders (Nephrogenic Diabetes Insipidus).

Dr. Bichet received the Medal of the Kidney Foundation of Canada in 1998, a Doctorat Honoris Causa from the University of Nancy (France) in 1999, and the Jean Hamburger Medal (the highest distinction of the European Society of Nephrology) in 2010.

Paul C. Grimm is a Professor of Pediatrics at Stanford University. He received his MD with distinction at the University of Saskatchewan, Canada in 1981. After serving as a rural family doctor in Northern Saskatchewan, he completed post-graduate training in pediatrics at The IWK Children's Hospital (Halifax), and Winnipeg Children's Hospital in 1986 and Pediatric Nephrology Fellowship at the IWK in 1988. He went to UCLA for further training in Transplantation Immunology, finishing in 1991. He returned to Canada, serving as Assistant Professor of Pediatrics at University of Manitoba until 1999, when he was recruited to UCSD as associate then full professor of Pediatrics and Medical Director of the Pediatric Kidney Transplant Program.

Recruited to Stanford in 2007, he has authored more than 60 peer-reviewed publications and is the recipient of numerous honors and awards, including a listing in "The Best Doctors in America" from 2002-2016. He serves as the Medical Director of the Stanford/LPCH Pediatric Kidney Transplant Program, Medical Director of the Cystinosis Referral Clinic and Director of the Pediatric Nephrology Fellowship Program. He is an appointed Member of the American Board of Pediatrics Subboard of Pediatric Nephrology, an Associate Editor of the Journals "Pediatric Transplantation," and "American Journal of Transplantation." He is a Councilor of the International Pediatric Transplant Association and a member of the Medical Advisory Board of the Cystinosis Research Foundation.

CRF-FUNDED RESEARCHERS HAVE PUBLISHED 63 STUDIES,

including these since the Fall 2015 issue of Cystinosis Magazine!

Hematopoietic Stem Cells Transplantation Can Normalize Thyroid Function in a Cystinosis Mouse Model

Published April 2016 online *http://press.endocrine.org/journal/endo*

by Pierre Courtoy, MD, PhD

DE DUVE INSTITUTE AND UNIVERSITÉ CATHOLIQUE DE LOUVAIN, BRUSSELS, BELGIUM

Impaired Lysosomal Function Underlies Monoclonal Light Chain-Associated Renal Fanconi Syndrome

Published March 2016 in *Journal of the American Society of Nephrology, JASN*

by Olivier Devuyst, MD, PhD

INSTITUTE OF PHYSIOLOGY, ZURICH CENTER FOR INTEGRATIVE HUMAN PHYSIOLOGY, UNIVERSITY OF ZURICH, ZURICH, SWITZERLAND AND DIVISION OF NEPHROLOGY, UCL MEDICAL SCHOOL, BRUSSELS, BELGIUM

Activation of the Transcription Factor EB Rescures Lysosomal Abnormalities in Cystinotic Kidney Cells

Published March 2016 online *http://www.kidney-international.org*

by Francesco Emma, MD, Division of Nephrology and Dialysis

BAMBINO GESÙ CHILDREN'S HOSPITAL AND RESEARCH INSTITUTE, ROME, ITALY

The AP-3 Adaptor Complex Mediates Sorting of Yeast and Mammalian PQ-Loop-Family Basic Amino Acid Transporters to the Vacuolar/Lysosomal Membrane

Published in *Scientific Reports*

by Bruno André, PhD

UNIVERSITY PARIS DESCARTES, FRANCE

Muscle Wasting and Adipose Tissue Browning in Infantile Nephropathic Cystinosis

Published in *Journal of Cachexia, Sarcopenia and Muscle*

by Robert Mak, MD, PhD

UNIVERSITY OF CALIFORNIA, SAN DIEGO

CRF TOTAL LEVERAGED GRANTS: $7,014,460

STÉPHANIE CHERQUI, PhD

1/1/2016 – 12/31/2020
RO1 NIH/NIDDKD:
Lentiviral-transduced hematopoietic stem cell transplantation for cystinosis.
Grant award: $1,395,000

THE IMPACT OF CRF

Stéphanie Cherqui was awarded a renewal of her "Lentiviral-transduced hematopoietic stem cell transplantation for cystinosis" grant! The total amount of the four-year grant award is $1,395,000. It will cover the impact of the large deletion 57kb on the stem cell treatment as well as other aspects that will improve the therapy.

This is extraordinary news for Stéphanie and for the cystinosis community. We are very excited for her and for the future of this research. Stéphanie has been the one who has said consistently for years that she would never give up until she found the cure for cystinosis. We have always believed in her brilliance and dedication to CRF and to our children.

Stéphanie has been funded by CRF since 2007. She formed the *Cystinosis Stem Cell and Gene Therapy Consortium* which includes experts in the fields of nephrology, neurology, endocrinology, gastroenterology, ophthalmology, bone marrow transplantation and gene therapy, and members of the cystinosis community.

CRF ISSUED $1,581,820 FOR BENCH, CLINICAL AND TRANSLATIONAL RESEARCH

In 2015 CRF issued ***9 new grants*** *that bring us closer to better treatments and a cure.*

LEPTIN SIGNALING IN INFANTILE NEPHROPATHIC CYSTINOSIS (INC)

Robert Mak, MD, PhD, ***Principal Investigator***
University of California, San Diego
$299,959 – 2-year grant
(February 1, 2015 – January 30, 2017)

Robert Mak, MD, PhD, at the University of California, San Diego, received a grant to study the underlying mechanisms of muscle-wasting in patients with cystinosis. The results of this study will pave the way for a novel therapy to treat muscle-wasting, a devastating complication from cystinosis that is associated with swallowing difficulty and respiratory weakness and the loss of mobility.

DEVELOPMENT OF TRANSDERMAL CYSTEAMINE DELIVERY SYSTEM

Ghanashyam Acharya, PhD, ***Principal Investigator***
Baylor College of Medicine, Houston, Texas
$112,500 – 1-year grant
(September 1, 2015 – August 31, 2016)

Ghanashyam Acharya, PhD, at Baylor College of Medicine in Houston, Texas, received a grant for the development of a cysteamine-infused transdermal patch, a potential novel treatment for cystinosis. A patch would eliminate the need to take dozens of pills each day and help patients who have difficulty swallowing.

NEWBORN SCREENING FOR CYSTINOSIS

Sihoun Hahn, MD, PhD, ***Principal Investigator***
Seattle Children's Hospital, Seattle, Washington
$153,929 – 1-year grant
(September 1, 2015 – August 31, 2016)

Sihoun Hahn, MD, PhD, at Seattle Children's Hospital, received a grant for the first newborn screening study for cystinosis, which, if successful, will save lives.

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

Stéphanie Cherqui, PhD, ***Principal Investigator***
University of California, San Diego
$364,800 – 2-year grant
(September 1, 2015 – August 31, 2017)

Stéphanie Cherqui, PhD, at the University of California, San Diego, received a grant for her cutting-edge bone marrow stem cell-mediated therapy.

MEDICAL DEVICE FOR STEM CELL AND GENE THERAPY STUDY - VIVASCOPE 3000

Stéphanie Cherqui, PhD, ***Principal Investigator***
University of California, San Diego – $82,583

Stéphanie Cherqui, PhD, at the University of California, San Diego, received a grant for a Vivascope which is a medical device to measure cystine crystals through the skin for the Stem Cell and Gene Therapy Study.

TRANSLATIONAL APPROACHES TO REPAIR CHAPERONE MEDIATED AUTOPHAGY IN CYSTINOSIS

Sergio Catz, PhD, ***Research Mentor***
Jing He, PhD, ***Research Fellow***
The Scripps Research Institute, La Jolla, California
$150,000 – 2-year grant
(February 1, 2016 – January 31, 2018)

Sergio Catz, PhD, at the Scripps Research Institute, La Jolla, California, was awarded a grant to continue his study on autophagy in cystinosis.

MOLECULAR MECHANISM OF CYSTINOSIS

Liang Feng, PhD, ***Mentor*** **+ Xue Guo, PhD,** ***Fellow***
Stanford University, Palo Alto, California
$225,000 – 3-year grant
(September 1, 2015 – August 31, 2018)

Liang Feng, PhD and Xue Guo, PhD, at Stanford University, Palo Alto, California, received a grant to better understand the molecular mechanism of cystinosis in an effort to learn more about the cause of cystinosis and to find the cure.

A STUDY ON BONE REMODELING DEFECTS IN NEPHROPATHIC CYSTINOSIS

Anna Taranta, PhD, ***Principal Investigator***
Andrea Del Fattore, PhD, ***Co-Principal Investigator***
Bambino Gesù Children's Hospital, Rome, Italy
$166,650 – 2-year grant
(September 1, 2015 – August 31, 2017)

Anna Taranta, PhD, at Bambino Gesù Children's Hospital in Rome, Italy, was awarded a grant to study the factors that contribute to bone remodeling defects in cystinosis.

USING YEAST AS A MODEL TO ELUCIDATE THE MOLECULAR IDENTITY OF LYSOSOMAL CYSTEINE TRANSPORTERS

Bruno André, PhD, ***Principal Investigator***
ULB – Institut de Biologie et de Médecine Moléculaires, Gosselies, Belgium
$26,400 – 1-year grant
(February 1, 2015 – January 31, 2017)

Bruno Andre, at Brussels Free Universities of Belgium, was awarded a grant to study the yeast model to learn more about lysosomal cysteine transporters.

USING YEAST AS A MODEL TO ELUCIDATE THE MOLECULAR IDENTITY OF LYSOSOMAL CYSTEINE TRANSPORTERS

Bruno André, *Principal Investigator*
UNIVERSITÉ LIBRE DE BRUXELLES

OBJECTIVE/RATIONALE:

The objective of this one-year project is to determine the molecular identity of the previously described human lysosomal cysteine transporter. This identification will open several avenues for the study of cystinosin, to be addressed in follow-up studies. For instance, it will enable us to evaluate the potential cooperation of this protein with cystinosin in the regulation of the intralysosomal cystine/cysteine concentrations. Furthermore, as this transporter is inhibited by cysteamine, it will be possible to study a potential link between this protein and cysteamine therapy.

PROJECT DESCRIPTION:

To determine the molecular identity of the human lysosomal cysteine transporter, we will use a combined genetic and biochemical approach in a model organism: the yeast Saccharomyces cerevisiae. We thus propose to first characterize at the molecular level the yeast transporter(s) catalyzing cysteine uptake into the vacuole. We expect these vacuolar cysteine transporters to be conserved in mammalian lysosomes. Therefore, in a second step, we will identify the human orthologs of the identified yeast transporters and initiate their functional characterization. This should open the prospect of further studying their role in the context of normal and cystinotic cells.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

We hypothesize that the lysosomal cysteine transporter to be identified acts in tight cooperation with cystinosin to ensure proper homeostasis of the lysosome. The study of such cooperation might shed precious light on the as yet poorly understood cause(s) of the various anomalies (reduced ATP pool, redox imbalance, signs of apoptosis…) described in the cells of patients with cystinosis. Furthermore, the cysteine transporter is inhibited by cysteamine, suggesting a potential link with cysteamine therapy.

ANTICIPATED OUTCOME:

We expect to unravel the molecular identity of the vacuolar cysteine transporter(s) of yeast, and to identify and initiate the functional characterization of possible orthologs from human cells.

TRANSLATIONAL APPROACHES TO REPAIR CHAPERONE MEDIATED AUTOPHAGY IN CYSTINOSIS

Sergio D. Catz, PhD, *Mentor*
Jing He, *Fellow*
THE SCRIPPS RESEARCH INSTITUTE

OBJECTIVE/RATIONALE:

To maintain normal function, cells need to degrade abnormal or damaged proteins and recycle their amino acids. One way this is achieved is through a process called chaperone-mediated autophagy (CMA). During this process, chaperones select abnormal or damaged proteins, and target them to the lysosome, an enzyme containing compartment that digests the proteins and recycles the amino acids. Previously we found that the CMA pathway is impaired in cystinosis and cysteamine treatment cannot recover this defect. Here, we propose to rescue CMA in cystinotic cell as a strategy to improve treatment.

PROJECT DESCRIPTION:

Our collaborator has discovered drugs that enhance CMA and we will test these drugs in murine cystinosin- deficient cells and human proximal tubule cells (hPTCs) obtained from two cystinotic patients. We will check the CMA activities and behaviors of the proteins that are involved in CMA pathway after treating cells with the drugs. In addition, we will test the drugs in cystinosin knockout mice. We will feed the CMA enhancers with or without cysteamine to the cystinosin mice, and then analyze the CMA activity and cystine accumulation in mouse kidney, liver and brain.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

Despite the immense improvements cysteamine has contributed to the treatment of patients with cystinosis, it nonetheless does not cure the disease, suggesting that the cystine accumulation is not the only contributing factor. Previous studies from our laboratory uncovered a novel role for cystinosin, mediating CMA, which highlights that CMA impairment is an important contributor to the pathogenesis of cystinosis and underscores the need for complementary treatments to cysteamine therapy. The work proposed here is highly translational, we believe that these studies will help develop new therapies for the treatment of cystinosis.

ANTICIPATED OUTCOME:

Our preliminary studies have already identified one CMA enhancer that dramatically improves the cellular function of cystinotic cells, and we will further test this enhancer in different assays. We expect this enhancer to rescue CMA activity in cystinosin deficient cells, and anticipate that cystinosin knockout mice treated with the CMA enhancer plus cysteamine will have better recovery than those mice treated with just cysteamine.

ACTIVITIES It Takes A Village CALENDAR

Friday, June 3, 2016

5TH ANNUAL LOTS OF LOVE FOR LANDON CHARITY GOLF OUTING

Lots of Love for Landon, Landon Hartz,
Black Hawk Golf Course, Beaver Falls, Pennsylvania
lotsoflove4landonCRF@gmail.com

Saturday, August 13, 2016

HUDSON BAY 4 HANK GOLF TOURNAMENT

24 Hours for Hank, Henry Sturgis, The Links in Post Falls, Idaho
Contact Luke Buoy for information (509) 201-0021

Saturday, September 10, 2016

SWING, SHOOT & LIV GOLF CLASSIC

Olivia Little, Liv-A-Little Foundation, Saugeen Golf Club, Port Elgin, Ontario
erin.little@livalittlefoundation.com

Saturday, September 10, 2016

5TH ANNUAL FORE FATHER'S JCFG MEMORIAL GOLF TOURNAMENT

Andrew Cunningham, Boulder Creek Golf Course, Langdon, Alberta
www.facebook.com/JCFGMemorialgolf

Saturday, September 24, 2016

3RD ANNUAL HEARTS FOR HADLEY EVENT

Hadley Alexander, Zion Bank Building, Boise, Idaho
hearts4hadley@gmail.com

Monday, October 17, 2016

CYSTINOSIS RESEARCH FOUNDATION NATALIE'S WISH NINTH ANNUAL FORE A CURE GOLF TOURNAMENT

Pelican Hill Golf Course, Newport Beach, California
For information contact Zoe Solsby, zsolsby@cystinosisresearch.org

Friday, December 16, 2016

STAR WARS PRIVATE SCREENING OF ROGUE ONE

Sam & Lars Hope for a Cure, Sam and Lars Jenkins
Sugar House Cinemark, Salt Lake City, Utah

March 2017

24 HOURS OF SCHWEITZER SKI EVENT

Henry Sturgis, Schweitzer Mountain, Sand Point, Idaho
www.24hoursforhank.org

Thursday, March 30 — Saturday, April 1, 2017

CRF **DAY OF HOPE** FAMILY CONFERENCE

The Island Hotel, Newport Beach, California
For information, contact Nancy Stack, nstack@cystinosisresearch.org

Saturday, April 1, 2017

CRF **NATALIE'S WISH** CELEBRATION

The Island Hotel, Newport Beach, California
For information contact Zoe Solsby, zsolsby@cystinosisresearch.org

LOOKING AHEAD

CRF Is Excited To Fund New Grants
IN PARTNERSHIP WITH RESEARCHERS AND SCIENTISTS AROUND THE WORLD!

In 2003, Nancy and Jeff Stack established the non-profit organization, Cystinosis Research Foundation (CRF), with the goal of funding cystinosis research to find better treatments and a cure for the disease. Since its inception, CRF has raised more than $33 million, with every dollar raised 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 early September CRF will announce the 2016 fall call for research proposals and fellowship grants. Details and guidelines for applications will be available online at the CRF website: w*ww.cystinosisresearch.org/research/for-researchers*.

Recently CRF received applications from the 2016 spring call for research proposals and fellowships. In evaluating the proposals, CRF utilizes a Scientific Review Board (SRB) comprised of leading international experts in the field of cystinosis (See page 70). 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. The SRB is currently evaluating the spring applications and CRF will announce the grant award recipients in July.

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 519 registrants from 43 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.

CYSTINOSIS RESEARCH FOUNDATION
RESEARCH • HOPE • CURE

Zoe R. Solsby
Vice President

MISSION

The mission of the Cystinosis Research Foundation is to find better treatments and a cure for cystinosis by supporting bench, clinical and translational research. Since 2003, CRF has raised $33 million for cystinosis research in an effort to find a cure.

EDUCATION

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


CYSTINOSIS
COMMUNITY
It Takes
A
Village
CYSTINOSIS
RESEARCH FOUNDATION
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HOPE
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