

# cystinosis

magazine

For friends and supporters of the Cystinosis Research Foundation



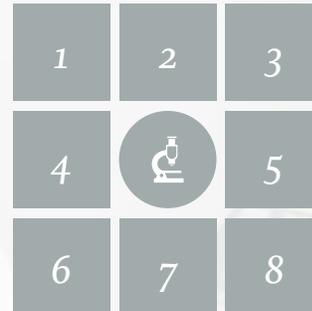
FALL 2015

HOPE THROUGH  
♥  
**RESEARCH**  
ISSUE



**ON THE COVER:**

8 researchers at the forefront of curing cystinosis.



1. Jennifer Simpson, MD
2. Francesco Emma, MD
3. Corinne Antignac, MD, PhD
4. Ghanashyam Acharya, PhD
5. Sergio Catz, PhD
6. Doris Trauner, MD
7. Pierre Courtoy, MD, PhD
8. Stéphanie Cherqui, PhD

**CONTACT US:**

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To receive our e-newsletter, *Star Facts*, send your e-mail address to [zsolsby@cystinosisresearch.org](mailto:zsolsby@cystinosisresearch.org)

The entire cost of Cystinosis Magazine is underwritten by friends of the Cystinosis Research Foundation.

Art Direction and Printing: Idea Hall

Dr. Jennifer Simpson (above), along with Dr. Ghanashyam Acharya, are making great strides to ease the protocol of taking eye drops every hour for corneal cystinosis.

**CYSTINOSISRESEARCH.ORG**

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FALL 2015

**cystinosis**  
magazine

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*CRF-Funded Research Offers Hope to Others*

SINCE  
**2003**

**\$29.3**  
MILLION

has been funded and committed to cystinosis research through the Cystinosis Research Foundation

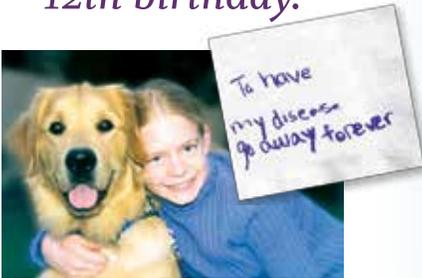


**58**  
ARTICLES

published from CRF-funded research!

+  
**100%**  
of the funds raised go DIRECTLY TO SUPPORT cystinosis research.

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



**THANK YOU**  
Your generosity has funded  
**131**  
MULTI-YEAR STUDIES  
in  
**12**  
C O U N T R I E S

+  
Your commitment has given new hope to  
**500**  
children and young adults living in the United States, and  
**2,000**  
cystinosis patients throughout the world.



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

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.



## A note from Natalie

Dear Friends and Family,

This past summer, I stayed in Los Angeles and interned at St. Anne's, a non-profit social service agency that specializes in serving the needs of at-risk pregnant and parenting young women and children. St. Anne's strives to empower and educate by providing care and resources to help these young women and children recognize their own value and potential, giving them hope for a better future. I had the opportunity to learn what goes on "behind the scenes" of a non-profit organization by working with the director of performance and quality improvement. I learned how important it is for all departments within the organization to work together to succeed. I thoroughly enjoyed working at St. Anne's.

In late August of this year, I began my second year of graduate school at the USC School of Social Work. This year, my internship is at the Office of the Federal Public Defender, where I am applying both my clinical and macro social work skills to real-life court cases. Although working on behalf of some of the offenders can be rather challenging, my experiences have given me a different perspective and attitude towards those who have been incarcerated. The accused are not always guilty, and it is important to give every human being a second chance in life.

My internships over the past couple of years have really helped me figure out what I want to do with my MSW degree when I graduate in May 2016. I have always had a passion to work with children and families and, recently, I have developed an interest in the public child welfare system. Eventually, I would like to go into family law or work at a family service, non-profit organization as a development director. I am continually growing both personally as well as professionally. I am looking forward to my future as a professional social worker.

As I continue to advance my degree and education, I realize how fortunate I am to have the cystinosis community support me. If it wasn't for our amazing community, I would not be as confident as I am today. With your support and dedication to finding a cure, I know I have a future. I know I will live a long life because of the tremendous research progress the doctors have made; I truly believe they will find a cure for this life-threatening disease. I am certain I will be able to accomplish my goals and aspirations because of the love, support, and encouragement I have received from the cystinosis community.

With  
your support  
and dedication  
to finding a cure,  
I know I have a  
future.

Thank you for your love and support,  
Natalie



NOVEMBER  
**2015**

### *Dear Family and Friends,*

As we approach the holiday season, we are filled with a sense of gratitude and thanks. As a result of your ongoing support, we are funding cutting-edge research at leading institutions around the world. CRF-funded researchers are forging ahead with bold ideas for innovative treatments and are always in pursuit of the ultimate goal – a cure for cystinosis.

### *You are Funding HOPE*

We have achieved research milestones because of the dedication of our outstanding group of CRF researchers, scientists, and clinicians who have dedicated their careers to finding better treatments and a cure for cystinosis. The cover of this issue of *Cystinosis Magazine* honors several CRF researchers who have made remarkable discoveries about the pathogenesis of cystinosis and novel ways to treat cystinosis, and have added to the overall breadth of knowledge in the field. As a result of their research, the cystinosis community has an abundance of hope that our children will live a life free of the disease.

### *The Realities of Cystinosis*

CRF-funded researchers have made new discoveries, and we have an improved medication. However, cystinosis remains an incurable, progressive disease. We are close to the cure, but we are not there yet. Between May and August of this year, our community mourned the loss of four young adults ages 23, 24, 25, and 31. In a community of only 2,000 people worldwide, the loss of so many in such a short period of time was devastating. Cystinosis destroys the organs in the body, including the kidneys, eyes, liver, muscle and brain. As our children reach adulthood, their symptoms increase, and their bodies deteriorate. The most severe complication for adults is muscle wasting, myopathy, and the progression of corneal cystinosis. We are renewing our efforts to fund research in these areas. The premature deaths of these young adults from our community are a reminder that we need to drive the research forward and find a cure for this devastating disease.

## Research at the Forefront

Since 2003, the year Natalie made her birthday wish to have her disease go away forever, and the year the foundation began, we have created a thriving research community. CRF aggressively and strategically invests your donations, issuing grants to the best and brightest researchers in the world. Initially, our research strategy was to fund basic or bench research. As discoveries were made in the lab, we funded clinical research, and now we are translating that research data into new treatments. In this magazine, you will find an illustrative summary of the research we have funded, the areas we have targeted, and the progress we have made (page 62).

### YOUR DONATIONS HAVE CHANGED THE COURSE OF CYSTINOSIS

CRF research milestones include:

- the first donor stem cell transplantation trial at UCLA,
- funding every bench and clinical trial that led to the discovery of a delayed-release medication that was approved by the FDA in 2013,
- new discoveries about the pathogenesis of cystinosis,
- and potential novel treatments.

When we talk to our children about the future, we talk about hope. The research we fund gives us hope that a cure will be found.

### More Than \$1.4 Million in New Grants

We are pleased to announce that seven new grants were awarded in July. Robert Mak, MD, at UCSD was awarded a two-year 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, weakness, and the consequent loss of mobility.

Ghanashyam Acharya, PhD, at Baylor College of Medicine received a grant for the development of a potential innovative treatment for cystinosis, a cysteamine-infused transdermal patch. We are ecstatic to fund Sihoun Hahn, PhD, at Seattle Children's Hospital for the first newborn-screening study for cystinosis that, when implemented, will save lives. CRF issued a new grant to Stéphanie Cherqui, PhD, for her cutting-edge bone marrow stem cell-mediated therapy at UCSD, as well as the acquisition of new lab equipment. CRF also funded a grant to Liang Feng, PhD, and Xue Guo, PhD, at Stanford University. Their study aims

to understand the molecular mechanism of cystinosis, and to learn more about the cause of cystinosis and the cure. Internationally, Anna Taranta, PhD, at Bambino Gesù Children's Hospital in Rome was awarded a grant to study the factors that contribute to bone-remodeling defects in cystinosis. We will announce another round of grants in December.

CRF has issued 131 multi-year research grants in 12 countries, and those researchers have published 58 articles in prestigious journals. CRF "seed" money has been leveraged by six NIH and other grants totaling more than \$5.6 million to two CRF-funded researchers. Together, we have funded researchers whose work has exponentially increased the breadth of knowledge about cystinosis. As a result of that knowledge, we are closer to the cure. The operating costs of the CRF are privately underwritten so that 100% of every dollar donated goes to research. CRF is the largest fund provider of cystinosis research in the world.

### Nanotechnology and Corneal Cystinosis

We continue to make progress on the nanotechnology project for corneal cystinosis. Corneal cystinosis is the painful eye condition that causes severe photophobia and sometimes blindness as our children reach adulthood. Fortunately, we have an eye drop treatment for corneal cystinosis, but it demands a rigorous protocol. The eye drops must be taken every waking hour in order to be effective. Compliance is an issue because the protocol is demanding, and the eye drops can be painful. Dr. Ghanashyam Acharya at the Baylor College of Medicine and Dr. Jennifer Simpson at UC Irvine have collaborated on research aimed at finding a better treatment.

Dr. Acharya invented a nanowafer that may revolutionize the treatment of corneal cystinosis. The concept is to place the nanowafer in the eye and, as it dissolves, the medication from the nanowafer stays in the eye, treating it for hours. CRF has obtained the license for the cysteamine-loaded wafer. This is a multi-year project, but we have an excellent team in place to ensure that we will reach our goal.

### Stem Cell and Gene Therapy

There is more good news from Dr. Stéphanie Cherqui's lab at UCSD. After some additional research and experiments using stem cell transplantation, Dr. Cherqui reversed corneal cystinosis in lab mice. Her article "Treatment of inherited eye defects by systemic hematopoietic stem cell transplantation" was recently accepted for publication in the IOVS (Investigative Ophthalmology and Visual Science)



**OUR FAITH IS STRONG  
AND OUR DETERMINATION  
STEADFAST, AND WITH ALL  
OF YOU BY OUR SIDE, THERE  
IS NO DOUBT WE WILL  
FIND THE CURE FOR  
OUR CHILDREN.**

Journal. She continues to work closely with the FDA to get final approval for the clinical study, which she anticipates she will receive by the end of 2016.

This year, Dr. Cherqui formed the Stem Cell and Gene Therapy Consortium that includes experts in the field of nephrology, neurology, endocrinology, gastroenterology, ophthalmology, bone marrow transplantation and gene therapy. She recently was awarded a grant from the Sanford Stem Cell Clinic that will cover the cost for the stem cell treatment of the two first patients enrolled in the upcoming clinical trial.

We are hopeful and optimistic that we will have the first autologous stem cell trial by the end of 2016! If this treatment works, it could *cure* cystinosis.

### ***Cystinosis Research Helps Others***

Discoveries made by CRF-funded researchers and scientists are currently being applied to other more prevalent and well-known disorders and diseases. There is great potential that CRF-funded stem cell research will help other corneal diseases, kidney diseases, neurological diseases, and genetic diseases with systemic defects similar to cystinosis. Your support of cystinosis research has extended far beyond the cystinosis community.

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

We are grateful to be surrounded by all of you who have embraced our children and our community. We thank you for giving so much of your time, energy, and resources to help us find a cure for cystinosis. It has taken all of us working together for our common goal of finding a cure for cystinosis to ensure research moves forward. CRF is the only foundation in the world funding millions of dollars in new research grants every year. We are the lifeline that keeps the cycle of cystinosis research community dynamic and thriving – without your support, we would not have hope.

### ***Thank You for Being Part of the Cure***

In this issue of *Cystinosis Magazine*, you will get to know more about our families, donors, and researchers. You will read about their commitment, determination, and common thread of hope and optimism.

Now more than ever, we are in a race to save our children. What I pray for every day is that Natalie and all other children and adults with cystinosis will have a life free of the disease – no medications, no pain, no muscle weakness, no hospital visits and blood draws, and no worries about their 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 you by our side, there is no doubt we will find the cure for our children.

We are within reach of new treatments and a possible cure that will change the course of cystinosis forever. With your support, we will continue to fund the best and brightest researchers in the world. Thank you for supporting cystinosis research, for standing by our side and embracing our community.

*With heartfelt thanks,  
Nancy and Jeff*

# 2016 Day of Hope

## CYSTINOSIS RESEARCH FOUNDATION FAMILY CONFERENCE

The Island Hotel, Newport Beach, California

Thursday, April 7  
— thru —  
Saturday, April 9  
2016



learn | share | laugh | celebrate

*The Day of Hope Conference and Natalie's Wish events are quickly approaching!*

This year's conference program will be very informative and provide a fun-filled weekend for the entire family.

In 2003 Natalie made her wish, "to have my disease go away forever." We know that many people with cystinosis and their families have that same wish.

We hope you and your family can join us for these very special events!

### CONFERENCE SCHEDULE

#### Thursday, April 7

Check-in at The Island Hotel  
Welcome reception and family dinner

#### Saturday, April 9

Conference sessions, family lunch  
and social time

#### Friday, April 8

Conference sessions, followed by  
family dinner celebration

#### Saturday Evening

Plan to join us for the exciting  
Natalie's Wish Celebration  
*(details on back cover)*

# BIG STEP FOR TINY WAFER

## Dr. Acharya Readies Promising Corneal Cystinosis Research for Clinical Trials

By Dennis Arp

**F**or a researcher who spends scores of hours in the lab each week chasing a much-anticipated medical breakthrough, perhaps no words resonate more powerfully than these:

“The results outperformed our expectation.”

Maybe that’s why Dr. Ghanashyam Acharya was smiling so broadly when he used those words to describe his latest testing of a tiny wafer that carries so much hope for patients with corneal cystinosis.

“I was thinking that – even if we achieved the same efficacy as with the current drug-delivery system – I would be fine with it,” said Dr. Acharya, a senior researcher specializing in nanomedicine at Baylor College of Medicine in Houston.

Instead, in testing with a mouse model using the nanowafer to deliver the medication cysteamine, the new system proved twice as effective with half of the drug concentration. More significant, the nanowafer heightened the stability and portability of the drug.

“The potential is great to improve treatment and compliance,” Dr. Acharya added.

As Dr. Acharya prepares an application to launch clinical trials of the nanowafer, which could revolutionize the treatment of cystine crystals in the eyes, he and his team are working on developing a transdermal patch that could greatly improve the lives of cystinosis patients who have difficulty swallowing.

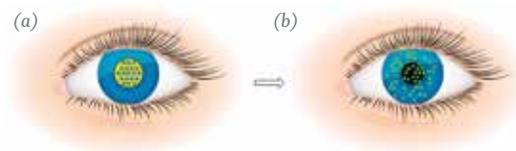
Both transformational projects are made possible by funding from the Cystinosis Research Foundation (CRF).

“Dr. Acharya is a brilliant engineer and a true advocate who is always thinking of new ways to help our community,” said Nancy Stack, president of the Cystinosis Research Foundation. “These projects hold tremendous promise for cystinosis patients and their families.”

The effort to develop a patch for delivering cysteamine through the skin is just beginning. However, with the nanowafer, Dr. Acharya and his team will apply to the Food and Drug Administration in 2016 for approval to begin clinical trials. If all goes well, those trials could begin late next year, with patients being enrolled at UC Irvine, Texas Medical Center in Houston, and possibly a third site.

### OCULAR DRUG DELIVERY NANOWAFER

(a) Schematic of the nanowafer instilled on the cornea.  
(b) Diffusion of drug molecules into the corneal tissue.



These are big developments for a tiny drug-delivery system that had no connection to cystinosis or ocular medicine until three years ago.

In March 2012, Dr. Acharya was recruited to the worldwide effort to seek treatments for patients with cystinosis by Dr. Jennifer Simpson, a pioneering researcher at the Gavin Herbert Eye Institute at UC Irvine. She saw the enormous potential for a big advance in cystinosis care offered by the nanowafer drug-delivery system.

Dr. Acharya quickly saw it, too. He learned that many cystinosis patients suffer from a painful buildup of cystine crystals in their eyes and on their corneas. Cysteamine drops help clear the crystals, but they must be reapplied hourly. Making matters more complicated, cysteamine drops must be shipped frozen and – once a bottle of drops is opened – the drops degrade so quickly that they must be discarded after a week.

Dr. Acharya speculated: what if he could pack a nanowafer with medication that would release slowly and continuously as the wafer dissolved in the eye? A single wafer might clear crystals for up to a week.

In tests with the mice, the results could scarcely be better. Not only was the drug-delivery system highly effective, but it made treatment dramatically more convenient.



Ghanashyam Acharya, PhD  
Baylor College of Medicine  
Houston, Texas



## TRANSDERMAL PATCH HOLDS TRANSFORMATIONAL PROMISE

For cystinosis patients who struggle to tolerate cysteamine pills, a transdermal delivery system will be a great leap forward. That's why Dr. Ghanashyam Acharya will launch a new project to test the effectiveness of a skin patch to deliver cysteamine.

A grant from the Cystinosis Research Foundation is funding the research.

Common side effects of cysteamine pills include vomiting, abdominal pain, diarrhea and loss of appetite. In addition, adults with cystinosis often have difficulty swallowing pills.

Dr. Acharya and his team will start by fabricating patches with various amounts of cysteamine to find a composition that enables an adequate dose to diffuse through the skin. Once they get the dosing right using silicone membranes and human cadaver skin, the expectation is that testing will move to clinical trials.

Ultimately, the researchers hope that their work will yield a noninvasive, disposable, inexpensive and highly effective alternative to cysteamine pills.

## “THE RESULTS OUTPERFORMED OUR EXPECTATION.”

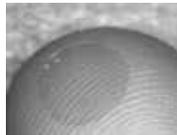
Dr. Acharya's testing showed that the medication in the nanowafer remained stable for up to six months without refrigeration.

“You can carry it in your bag, like a Band-Aid,” Dr. Acharya noted.

Ultimately, that ease of storage should help reduce the cost of treatment. With increased competition from pharma companies, cysteamine drops likely will become more affordable as well.

CRF owns the licensing rights to the nanowafer with cysteamine – a reflection of Dr. Acharya's commitment to cystinosis patients and the organization.

“We are setting up a new thinking process,” Dr. Acharya said. “I'm very grateful for the CRF's steadfast support, and it can administrate things so treatment will be better for patients. The treatment is more important than profits.”



*Nanowafer  
on a fingertip.*

Dr. Acharya's connection to cystinosis patients and the CRF has deepened during visits to the Day of Hope conference during the past two years.

“When you go there, you understand the sense of urgency,” he said of the chance to meet with other researchers and cystinosis families. “It's why we (researchers) work so late into the night.”

Dr. Acharya's work remains focused on the day when a medical advance becomes available to patients so it can improve their lives. With the nanowafer for cystinosis, he hopes that day will arrive as soon as a year from now.

“That's the satisfying moment we all want,” Dr. Acharya said. “That's what gives meaning to our daily lives.”

# TRANSPLANTING HOPE

*Dr. Stéphanie Cherqui Takes the Next Step  
in Her Innovative Genetic Research*

By Dennis Arp

When one's research shows as much promise as Dr. Stéphanie Cherqui's, it's natural for patients to think ahead to when a potential breakthrough will make their lives better. Dr. Cherqui empathizes, but she understands that breakthroughs involve a process with clear steps that lead to success.

"I know that families are eager to see our clinical trial start, and we are, too," said Dr. Cherqui, whose research with a mouse model has shown that transplanting stem cells from a matched related donor can slow or even prevent the progression of cystinosis.

Her groundbreaking pathway to a possible cure is funded by a grant from the Cystinosis Research Foundation and by the National Institutes of Health.

"We're doing everything we can to be on time (with our studies), as we want to guarantee that every safety step is taken," she added.

Dr. Cherqui and her team at the University of California, San Diego, are in the third year of an investigation to pursue approval by the Food and Drug Administration (FDA) for their gene therapy approach. She and her colleagues are now trying to show that a patient's own stem cells can be altered to genetically express a functional cystinosis (CTNS) gene.

It's all part of the heroic effort to correct the defective gene that is the basis for cystinosis.

"Our gene therapy is considered to be a new drug, so we have to submit a new IND (Investigational New Drug) application," Dr. Cherqui noted. "We hope to submit it in about a year, then we'll have to see if they're OK with all we've done."

Because there are so many variables, she hesitates to predict outcomes or an end point to her investigation.

"I always warn that – although this treatment has worked great in mice – there are no guarantees it will work in humans," Dr. Cherqui noted.

Still, her research with cystinosis patients remains powerfully encouraging. Nine patients have provided blood for studies. When the official clinical study begins, six patients will be initially involved: four of them will be 18 or older, and two will be 13 or older.

If the research realizes its promise, it could usher in "a new era for treating multisystemic lysosomal-storage disorders," according to Dr. Cherqui.

In addition to cystinosis, other diseases are already in clinical trials using this stem-cell gene therapy approach and have realized successful outcomes, including

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**IT COULD  
USHER IN "A  
NEW ERA FOR  
TREATING  
MULTISYSTEMIC  
LYSOSOMAL-  
STORAGE  
DISORDERS."**

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blood disorders, cancers and those characterized by immune deficiencies.

While everyone working on the genetic research does so selflessly, Dr. Cherqui is quick to point out that the focus should remain squarely on cystinosis patients.

"The first research volunteers are pioneers in the process," she said. "When I talk with them, they ask amazing questions. You can see that they've thought through the whole process. I'm impressed to see the maturity demonstrated by all these patients."



Stéphanie Cherqui, PhD  
University of California, San Diego  
San Diego, California



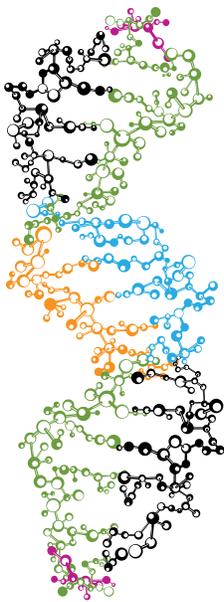
## GRANT AWARDS ARE LEVERAGED BY MILLIONS \$5,619,460

STÉPHANIE CHERQUI, PhD

- 1/1/2011 - 12/31/2015  
R01 NIH/NIDDK:  
**Lentiviral-transduced hematopoietic stem cell transplantation for cystinosis.**  
Grant award: \$1,445,615.
- 8/6/2013 - 5/31/2015  
R01 NIH/NIDDK:  
**Toxicology studies for gene-modified stem cell transplantation for cystinosis.**  
Grant award: \$539,400.
- 9/1/2011 - 8/31/2013  
NIH/NIDDK R21-DK090548-01A1:  
**Kidney-targeted gene delivery for cystinosis.** Grant award: \$473,750.
- 8/15/2014 - 7/31/2016  
R21 NIH/NINDS:  
**Hematopoietic stem cell-based therapy for a neurodegenerative disorder.** Grant award: \$426,250.
- 9/1/2015 - 8/31/2017  
Sanford Stem Cell Clinical Center:  
**A one arm, open label, single treatment safety and efficacy study of pCCL-CTNS modified CD34+ hematopoietic stem cells after autologous transplantation in patients with nephropathic cystinosis.** Grant award: \$1,204,445.

JENNIFER SIMPSON, MD

- 4/2012 - 4/2016  
NIH R01 EY022365: **Mesenchymal Stem Cell Therapy for Corneal Cystinosis.** Grant award \$1,530,000.



## THE IMPACT OF CRF

Two CRF-funded researchers have collectively been awarded \$5,619,460 in additional grant funds by other institutions as a result of “seed” money provided by CRF. The discoveries made by CRF researchers benefit the greater scientific community and assist other more prevalent diseases and disorders. By funding the cystinosis research community, you are effectively helping millions of people with other diseases.

Those who sign onto the trial know it's a two-year commitment, during which time they will be closely monitored, especially at the time of stem-cell transplantation. That part alone means a month-long hospital stay plus weekly checkups for three months after transplantation.

“They go in realizing there may be no benefit (from the treatment), but they're ready to try anyway,” Dr. Cherqui said. “I admire that.”

# INSIDE STORY

## A Unique Study Will Provide a Deeper Look at Bone Development

By Dennis Arp

**F**or 17 years, Dr. Mary Leonard has studied how childhood chronic diseases affect the development of bones. Now, she's eager to get a look at cystinosis from the inside out.

Dr. Leonard and her colleagues at a new Stanford University research center are conducting a first-ever study using 3D imaging to assess bone structure in patients with cystinosis. The study is also the first to consider the relationship between muscle strength and bone structure.

The findings will help determine how cystinosis changes bones, what risk factors need special attention, and what therapies might make things better.

“As a community, we are desperate for information about these musculoskeletal issues,” said Nancy Stack, president of the Cystinosis Research Foundation (CRF), which is funding the research.

Children and adolescents with cystinosis harbor a daunting array of risk factors for poor bone development. Among them are

malnutrition, urinary phosphate wasting, hypothyroidism, muscle weakness, cystine crystals in the bone and consequences of chronic kidney disease.

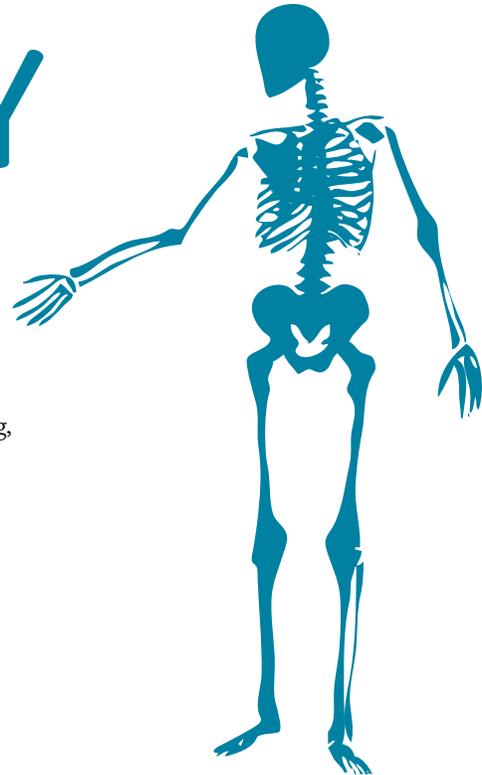
“If you don't develop good bone structure by your 20s, you can't make bones thicker or denser,” said Dr. Leonard, professor of pediatrics and associate dean of maternal and child health research at Stanford University. “By getting a broader and deeper look at the impairment, it means that we can see the source of this fragility.”

Dr. Leonard is partnering on the research with Dr. Paul Grimm, a Stanford clinician and nephrologist who is much-beloved for his work with cystinosis patients.

“I'm the bone expert, and he's the clinical cystinosis expert,” Dr. Leonard said.

The expected year-long study began in mid-September 2015, when the first two patients were seen. Researchers hope that 30 children and adults with cystinosis will enroll, with 60 more non-cystinosis patients participating as controls.

High-resolution, low-radiation cat (CT) scans will help assess bone density and structure in the forearm and lower leg. In addition, conventional bone density (DXA) scans of the whole body, hip, spine and forearm will provide measures for comparison with CT results.



DXA scans will help measure leg muscle mass, and exercise equipment will be used to assess leg strength.

The researchers have begun this research with several hypotheses. One is that phosphate losses in the urine and insufficient phosphate replacement through supplements are associated with low bone density. Another is that low muscle mass and poor muscle strength are linked to thin bones.

Whatever the study findings, according to Dr. Leonard, “the information will help physicians better understand risk factors for bone development, and it will lead to clinical trials that target these risk factors.”

In developing the study, the researchers have benefited from observing cystinosis patients and talking with their families – especially during the annual Day of Hope event sponsored by the CRF.

“Parents would come up to me during breaks and describe how their children





Mary Leonard, MD  
Stanford University School of Medicine  
Stanford, California  
+



## “CHRONIC KIDNEY DISEASE IS ALREADY ASSOCIATED WITH HIGH BONE FRACTURE RATES.”

have subtle abnormalities in their bones and joints,” said Dr. Leonard. “One parent told me, ‘Go over to the pool and watch the kids.’ I saw that, for some of them, their ankles are turned in. This helped me see the value in bringing a pediatric orthopedic surgeon onto the research team. I haven’t done this before, but I think it will be a nice precedent.”

Dr. Leonard notes that chronic kidney disease is already associated with high bone-fracture rates. With so many cystinosis patients on kidney dialysis or for those who have received a transplant, “we will need to tease apart the impact of cystinosis to separate it from the kidney disease,” she says. “That’s another layer of complexity to the study.”

As the researchers immerse themselves in the project, they feel supported by the “exceptionally impressive” enthusiasm of cystinosis families as well as the hopeful spirit of the patients.

“Cystinosis is perfect for this study,” Dr. Leonard added. “I feel very fortunate to be working with such a committed community.”

## VOLUNTEERS NEEDED

Volunteers are needed for Dr. Mary Leonard and Dr. Paul Grimm’s study on musculoskeletal disease in children and adults with cystinosis.

This clinical study at Stanford University is fully supported by a 2015 grant from CRF. Patients with cystinosis have numerous risk factors for impaired bone accrual, including myopathy. This is the first comprehensive evaluation of bone structure and muscle strength in children and adults with cystinosis. We hope the results of this trial will lead to new clinical trials to improve bone health, muscle strength and quality of life in patients with cystinosis.

### 30 cystinosis volunteers and 60 controls are needed for the study.

For more details, please contact Jessica Whalen at [jwhalen@stanford.edu](mailto:jwhalen@stanford.edu) or call her at (650) 683-5287. Thank you to cystinosis families, patients, and our friends and family for supporting this study. We thank the volunteers who will be part of this study - you will help us learn more about cystinosis so that we can target research aimed at better treatments and a cure.

See Ad on Page 56.

# BABY STEPS

## *Breakthrough Research Could Lead to the Screening of Newborns for Cystinosis*

By Dennis Arp

**F**or translational researcher Dr. Sihoun Hahn, professor of pediatrics and adjunct professor of medicine at the University of Washington and Children’s Hospital, and the director of biochemical and molecular genetics laboratory at the University of Washington, a career made of life-changing laboratory work has been built on one simple bromide.

Dr. Hahn’s research focuses on developing screening methods for rare diseases so patients can be treated even before they start showing symptoms.

“I’m a true believer in prevention,” said Dr. Hahn. “Identifying illness and providing interventions are always preferable to treating a developed disease.”

Thanks to a grant from the Cystinosis Research Foundation (CRF), Dr. Hahn is now working on an innovative tool to test newborn babies for cystinosis. The method also shows great potential for detecting many other congenital disorders.

The test would quickly and accurately screen for cystinosis using blood spots from newborns. A simple prick of the heel while a baby is still in the hospital nursery could be the first step to testing for several different disorders. Currently, newborns are tested soon after birth for more than 30 rare disorders.

For more than 15 years, Dr. Hahn has focused his research on population screening for Wilson’s disease, a genetic disorder in which the

body fails to excrete copper properly, leading to the metal’s accumulation in various organs, including the liver and brain.

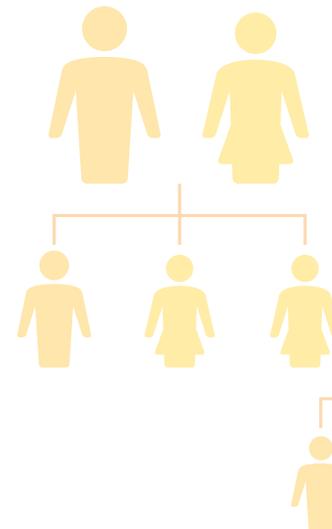
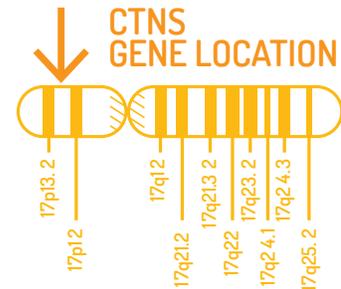
In performing this research, Dr. Hahn and his colleagues gained valuable insights into next-generation technology for genetic sequencing. That experience informed their more recent efforts to develop what amounts to a peptide fingerprint for cystinosis as part of next-generation newborn screening.

If the testing continues to work as well as it has so far, the innovative assay will give clinicians a test using spectrometry – a technique that helps identify the amount and type of peptides present in a dried blood spot sample.

“We have to test a lot of peptides with sophisticated equipment,” said Dr. Hahn. “Our ultimate goal is to automate about 15 steps in the process.”

That automation, which includes the use of robotic instruments, is important because, according to Dr. Hahn, researchers “have to run 200 or more samples each day, and you can’t do that manually.”

It will become even more critical to automate processes once the test moves beyond the experimental phase. In the state of Washington alone, more than 70,000 babies are born each year, and the hope is to develop a test that becomes a mandated newborn screening used throughout the nation and eventually the world.





Sihoun Hahn, MD, PhD  
Seattle Children's Hospital  
Seattle, Washington



## THE IMPORTANCE OF NEWBORN SCREENING

CRF is proud to fund Dr. Hahn's study on newborn screening for cystinosis. Newborn screening is a critical tool that will help us identify babies who have cystinosis so that they can begin cysteamine treatment immediately. In almost all cases of cystinosis, our babies look normal and healthy at birth. Diagnosing cystinosis soon after birth before the severe signs of the disease develop offers the best chance for successful and effective treatment. Not only does early treatment help growth and development, it helps protect the vital organs that are damaged early on without cysteamine treatment. Newborn screening will help save our children's lives.

---

Dr. Hahn's study is the first study ever funded on newborn screening and a crucial step toward the development of a population screening test that will have a high impact on cystinosis patients' outcomes.

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"Early initiation of oral cysteamine therapy prevents the progression of renal glomerular disease and enhances growth in nephropathic cystinosis; it also prevents late complications of the disorder. I have repeatedly made the point that the single most critical advance for the entire field of cystinosis would be a successful newborn screening program!"

---

*William A. Gahl, MD, PhD*

*Clinical Director,  
National Institutes of Health*

---

**"I'M A TRUE BELIEVER  
IN PREVENTION  
... PROVIDING  
INTERVENTIONS IS  
ALWAYS PREFERABLE  
TO TREATING  
DEVELOPED DISEASE."**

"We still need to run pilot samples, but we're optimistic," he said.  
"We have very positive findings."

The positives will multiply once a screening of infants becomes standard because, "when we start treatment in the newborn period, patients realize a much higher quality of life," Dr. Hahn noted. "Pre-symptomatic treatment – that's what we all want to see."

Realizing that dream may take years of diligent work, but the process definitely benefits from the support of cystinosis families and the CRF, Dr. Hahn added.

"Our supporters make this opportunity possible," he said.  
"We're blessed and humbled to have them as partners."

# Happy Holidays

## FROM THE CYSTINOSIS RESEARCH FOUNDATION

### *Dear Family and Friends,*

As we celebrate the holidays, what better time to thank all those who make the Cystinosis Research Foundation so exceptional and successful.

- *Thank you* to the children and adults with cystinosis who inspire us every day with their relentless battle against cystinosis.
- *Thank you* to family members of those with cystinosis who are always there to support their loved ones with cystinosis.
- *Thank you* to our friends and donors who steadfastly stand by us on the long and winding journey towards a cure.
- *Thank you* to the CRF funded scientists, researchers and clinicians worldwide who have dedicated their careers to our children and who work tirelessly on behalf of every child and adult with cystinosis.

Each one of you plays a critical role in the fight against cystinosis. We would not be where we are today without you. We have made great research progress — we won't stop until a cure is found. Thank you for giving us the gift of hope.

With blessings from our family to yours,

*Nancy and Jeff Stack*

To make a year-end gift to the Cystinosis Research Foundation,  
[www.cystinosisresearch.org/how-to-help/donate-online](http://www.cystinosisresearch.org/how-to-help/donate-online)



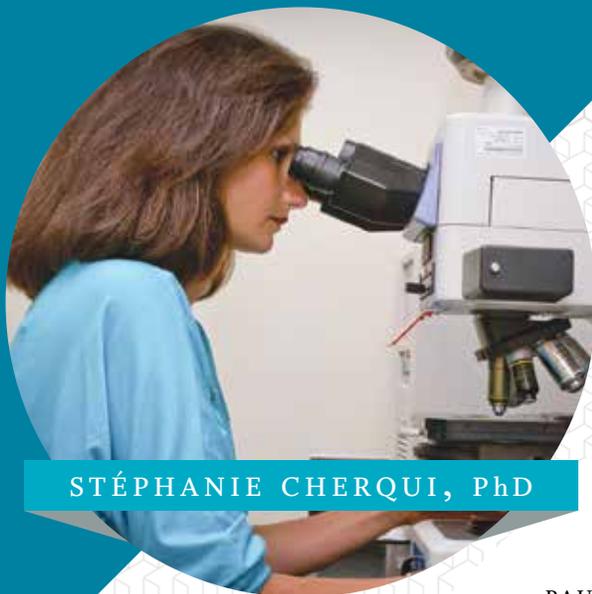
**CYSTINOSIS RESEARCH FOUNDATION**

[www.cystinosisresearch.org](http://www.cystinosisresearch.org) ★ 949-223-7610

The Cystinosis Research Foundation is a non-profit, tax-exempt entity pursuant to Section 501(c)3 Federal Tax ID #32-0067668

# A GIANT STEP TOWARDS THE CURE

**T**he Cystinosis Research Foundation (CRF) is excited to announce that we are one step closer to the cure. Dr. Stéphanie Cherqui, who has been funded by CRF since 2007, recently formed the **Cystinosis Stem Cell and Gene Therapy Consortium**. The consortium includes experts in the fields of nephrology, neurology, endocrinology, gastroenterology, ophthalmology, bone marrow transplantation and gene therapy, and members of the cystinosis community. Collectively, the consortium will contribute to the design and methodology of a clinical trial for cystinosis patients that will test the safety of autologous transplantation with hematopoietic stem cells that have been gene modified ex-vivo with a lentiviral vector to express a functional CTNS gene.



STÉPHANIE CHERQUI, PhD

The consortium members include:

STÉPHANIE CHERQUI, PhD  
*Study Director, UCSD*

EDWARD D. BALL, MD  
*Director of Bone Marrow Transplantation at UCSD*

NADINE BENADOR, MD, *Nephrology, UCSD*

BETTY CABRERA, *Clinical Trial Coordinator, UCSD*

RANJAN DOHIL, MD, *Gastroenterology, UCSD*

PAUL GRIMM, MD, *Nephrology, Stanford University*

ERIC NUDLEMAN, MD, *Ophthalmology, UCSD*

DONALD B. KOHN, MD, *Hematopoietic Stem Cell Gene Therapy, UCLA*

ROBERT MAK, MD, *Nephrology/Muscle, UCSD*

SUSAN PHILLIPS, MD, *Endocrinology, UCSD*

NANCY STACK, *President, Cystinosis Research Foundation*

DORIS A. TRAUNER, MD, *Neurology, UCSD*

# Liv-A-Little

By Erin Little, Olivia's mom

PORT ELGIN, ONTARIO, CANADA

Olivia will turn six years old on December 14. She is doing extremely well. She's growing slowly, but still growing. She is gaining weight, but that is slow going as well. She continues to maintain the 3rd percentile on the growth chart, but she keeps up with children in the 90th percentile on the growth chart, and for that we're grateful.

Olivia still has no idea that she is different from her friends or even her sister, which we consider to be a blessing. We know one day she'll connect the dots, and we'll have a lot of explaining to do. However, we remain hopeful that we'll have a cure before that day comes.

Summer has ended, so we packed Olivia's golf clubs and beach toys away. Olivia started golf lessons this summer. She loved her time on the course and loved her coach, Mr. Mike. She has also discovered a love for the water, so we spent a lot of time at the sandy beach. As soon as we put her floaties on, she would dash into the water and would not come out until her skin was wrinkled like a prune.

Golf and swimming were two sports we dreamed Olivia would like because they are sports that will help her build muscle, are gentle on her bones, allow her to stay cool, and she competes with herself (unless it's golf, then she's competing with us). But, most importantly, she has fun playing these sports. This was the first year Olivia has played a sport. The timing was perfect because she is more vocal and can express her two important needs: bathroom breaks and "more water, please."

Fall has been an exciting time for Olivia; she started her homeschooling adventure and is officially a kindergarten kid. Olivia is a very particular kid; many would say she is on the path of becoming a type A personality, as evidenced by her strong opinions when it came to setting up our school room. After I arranged the school room, I anticipated that she would see the room and fall in love with her beautiful classroom. Instead, she told me (with her newly famous pout face) that it wasn't



## PAINT THE TOWN PURPLE

Once again, the town of Saugeen Shores joined together to show its love and support for funding a cure for Olivia. The “Paint the Town Purple” campaign to raise awareness about cystinosis was nothing short of amazing! Purple bows popped up on local businesses, light posts and front doors of houses around town. The campaign generated interest from locals and tourists who, after hearing Olivia’s story, donated to help find the cure.

Scott Rowland, the owner of Rowland’s Independent grocery store, proudly displayed the purple bows. The store was filled with purple balloons and bows for two weeks, during which time Rowland’s Independent encouraged customers to donate to cystinosis research upon check out. This year, in just two short weeks, more than \$10,000 was raised. The entire Little family, including Olivia and her sister Harper, spent a few days bagging groceries to give back to those who were giving to cystinosis. Olivia had a wonderful time helping and continues to say that when she grows up, she wants to work for Mr. Scott! Saugeen Shores is an incredible community.

a classroom because there needed to be a desk and chair at the front of the room so that I could stand in front of the room at the chalkboard and teach her! My first thought was: “This is going to be a long 12 years!”

Along with starting kindergarten, she started ballet. Olivia loves to dance and, like any other little girl, she dreams of the costume. She proudly tells everyone about her ballet, and shares that all ballerinas need to earn their tutus. Watching her grow to be a beautiful little girl is the best gift.

The other major change in Olivia is her newfound independence. It is wonderful and heartbreaking at the same time. Her new phrase is, “I can do it myself,” which Harper, her younger sister, is also saying. It started when she prepared her own breakfast for the first time. Making her breakfast consisted of going to the fridge, grabbing whatever fruit she could reach, washing it and cutting it up. The other night, I shed a tear when she went upstairs and showered all by herself. She was so proud of herself when she got out and couldn’t wait to tell me all about how she used the soap, then laughed when she told me she forgot to wash her hair and how she would remember the next time.

Her independence expands daily. The most poignant example of her new independence was when she woke up from her afternoon rest, yelled from the top of the steps that she was awake and was going to play quietly in her room, then told me to come get her when it was time for her eye drops and vitamins. I am so proud of her. I knew at that moment that she would be okay, and I know for now that she’s on the right path to learning how to take care of herself. We pray that she continues on this path.

*Right now, cystinosis isn’t leading her life; she is. She is living in the moment, and cystinosis is along for the ride.*

TINA FLERCHINGER

# Tina's Heartfelt Words

By 12-year-old Tina Flerchinger and Denice Flerchinger, Tina's mom  
LEWISTON, IDAHO

**A Mother's Reflection:** *On this early fall morning, I am in flight to a Cystinosis Research Foundation board meeting. As I watch the sun rise on the horizon, I can't help but tear up. Twelve years ago today, God sent us the gift of another beautiful daughter. Our lives were changed that day. There have been a lot of bumps in the road, but we have come so far, and we are truly blessed. Following is an excerpt from Tina's journal.*

I'm in sixth grade now! I am turning 12 and getting my ears pierced. I am doing great in middle school, and I have four different teachers. School started for me four weeks ago, but this week was hard. I didn't feel so well.

Saturday morning, my mom woke me at 6:00 a.m. to give me my pills in bed and let me go back to sleep. I never get to sleep in; I was happy! When I woke up my sister Catherine made me my favorite turkey sandwich for breakfast. Then, I took more pills, because I can't take all 21 pills at the same time.

I was gagging so much I just knew it was going to come out and, sure enough, it did. I did not like throwing up because I knew I would have to retake my meds. And, every time I throw up, my meds usually come back out. Some of them get stuck in my throat; it hurts very badly. I take the rest of my pills, then I go upstairs and take a bath. When I am done taking a bath, I throw up again. I feel miserable, and I know I can't go to my cousin's wedding because I feel so sick. I tell mom, and she says I can stay home with Catherine.

My dad says I need something in my stomach and makes me an egg. Then my parents leave for the wedding, and so I watch some TV. I still don't feel good. I read a chapter in my book. Then, I cuddle with my puppy Sam while napping. When mom and dad get home, I tell them I still don't feel good. They can tell because I am always happy unless I do not feel good.

We eat dinner. My dad makes steak. Usually I love steak, but I wasn't really hungry. I ate a couple of bites, but that's all. As we were doing dishes, I suddenly started feeling worse. Every time I take my meds or eat, it comes up. My mom is worried about me. She sleeps downstairs on the couch, and I sleep on the other couch. I try to fall asleep, but I can't because I feel miserable. I throw up again for the 10th time today.

I am not doing well. Mom and Dad take me to the hospital. My dad leaves the car at the emergency room door and carries me in. The nurse puts a blood pressure cuff on my arm. We go into a different room, and I lay on the bed. My stomach is aching so badly. My stomach has been aching since school started. The doctor comes in and asks, "What's wrong?" My parents tell him everything. Then the doctor puts an IV with medicine in me. I feel better, but my stomach still aches. My stomach feels worse. We were at the hospital for three whole hours. I was just lying there, and I couldn't even fall asleep because of all the cords and stuff that were connected to me. Then, the doctor came in and talked to us; he recommended that I take more medicine. The nurse gives me two doses of another medication, and we head home.

When we get home, it is about 3:00 a.m. I am tired so I head upstairs. I brush my teeth, I shut my curtains, I turn the lights off,  
*I get in bed, and then I say my night prayers and thank Jesus that I am doing much better.*

“

*My mom is worried about me. She sleeps downstairs on the couch, and I sleep on the other couch. I try to fall asleep, but I can't because I feel miserable. I throw up again for the 10th time today.*

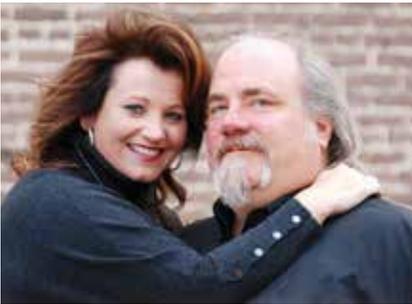
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## JULIE AND CLARK KLAPPENBACH PROVIDE WEDDING GIFTS OF HOPE

By Julie Klappenbach



**I COULDN'T BELIEVE THAT THIS BEAUTIFUL AND PERFECT EXAMPLE OF GOD'S CREATION, TINA, WAS ENGAGED IN SUCH A LIFE-THREATENING BATTLE.**

**W**hile visiting my parents in the Lewiston-Clarkston Valley several years ago, I attended Mass with my Dad. He introduced me to the Flerchinger family. He always sits two rows behind them, as if there were assigned seats. On our drive home from Mass that day, I remember Dad telling me about the disease that Tina is fighting. I couldn't believe that this beautiful and perfect example of God's creation, Tina, was engaged in such a life-threatening battle. How was it possible that this charming, young beauty could be struggling with this disease and continue to smile so radiantly? I immediately felt the urgency to help her family work to find a cure.

Since that time, I have relocated to Lewiston to be closer to my family. I get to see Tina's adorable smile each Sunday at Mass, and I have been honored to help during the annual event for TINA'S HOPE FOR A CURE: WINE, STEIN AND DINE. Working alongside Tina's family at this event is one of the things I look forward to each spring.

My involvement with the charity got me thinking about additional fundraising opportunities.

Part of today's wedding planning can involve building a "wedding website." My fiancé and I chose to build our wedding website via TheKnot.com. On the registry

page, TheKnot.com allows the marrying couple to pick a charity to add to the list. This function donates a small percentage of the amount spent on gifts purchased for the bride and groom through their registry page to a dedicated charity. The existing available list didn't include Tina's Hope for a Cure. And, a lightbulb went off for me!

I worked with Tina's mother, Denice, to figure out how to get Tina's Hope for a Cure listed on our registry page. Success!

Adding Tina's Hope to our Registry page made sense for us because Clark and I found each other in our forties. We each had a well-established inventory of household goods. Sure, we needed to add new towels and a few other items on our traditional registry, but we truly didn't "need" anything. For us, adding this charity to our wedding registry was perfect.

Denice kept us well informed of the donations that were submitted in our name. Each time I saw an email from her, I had to grin because I knew that we are that much closer to finding a cure for cystinosis and our young friend, Tina.

Clark and I are honored to have had Tina's Hope for a Cure listed on our wedding registry, and we are so pleased that some of our friends and family chose to honor our marriage by donating.

# DO YOU OR SOMEONE YOU LOVE HAVE CYSTINOSIS?

*Are they affected by:*

**Photophobia?**

**Eye Pain?**

**Foreign Body Sensation?**

**Squinting?**

If you said "yes" to any of the above, it may be time to discuss these symptoms with your ophthalmologist.

Cystinosis causes cystine build-up in the body which may damage cells in the kidneys, liver, brain, other organs and the corneas.



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GEN10 08/15

# “We’re OK. We’ll be OK.”

By Barbara Gruden,  
Ajda’s mom  
SLOVENIA

I can say those words now, and most days, I truly believe them. I’m Barbara, and I am married to Miha. We have two kids, Žiga is 3 (almost 4, he’s very accurate about that) and Ajda is almost 12 months. And, we have a dog named Benji. We live in Slovenia, on the sunny side of the Alps. We’re just a normal, everyday family. Well... not exactly.

We’ve had some hard months, which started early this year (March 20th, to be exact) on the day our nephrologist told us that our beautiful, healthy daughter Ajda has cystinosis. The memory of that moment is so alive, and yet it sometimes feels like a dream, a bad dream. But, it was not a dream. It was the beginning of our new life.

During the first month after Ajda’s diagnosis, we were just trying to put the pieces together. The second month (for me especially) was a month living in denial and feeling sorry for our situation. Then, during the third month, we started to build our lives again. We’re doing it slowly, step-by-step, with some bumps along the way.

In the beginning, it was quite hard when Ajda stopped eating and began to vomit – a side-effect from cystagon. I still remember the day when our nephrologist gently explained to me that Ajda might need a G-tube. “No way,” were my first words. Our world had just fallen apart, and now the doctors were telling me that our daughter needed a tube in her belly: “no way,” I thought.



## AJDA GRUDEN

A month later after struggling to get Ajda to eat, we changed our minds and begged the doctor to put the tube in as soon as possible. Before the G-tube, our days were centered around food. I would send everyone out of the room so Ajda could eat a bit. But in the end, she always vomited. We had to get our lives back, and getting the G-tube and accepting that change were the first steps.

By the time we finally got the G-tube, Ajda had lost more weight and was so skinny. She weighed the same at nine months as she did at four months. It was hard watching her not having any energy in her little body. A week after getting the G-tube, Ajda switched from Cystagon® to Procsybi®. And, guess what? She started eating again! When her Procsybi® dosage was low, she had a big appetite. But as her dosage increased, her appetite decreased marginally. Today, we're at the full dosage, and she eats well. We are grateful that the vomiting and sickness have stopped – at least for now.

With the support of our family and friends, we are getting our lives back. We are trying to live as normally as we can, enjoying the good days and trying to survive the bad days. We couldn't get through the really bad days without our children's smiles. No matter what, Ajda is always smiling, and Žiga is such an amazing child and a terrific big brother.

During the hard times, it helped so much to have the comfort and support of our new friends in the U.S. Some friends we met at the beginning, some along the way, and we are grateful for all of them. I know we are not alone, and I know that every time I have a question or get spooked about something, support and comfort is just an email away.

I still have my dark days, when all those scary thoughts come rushing into my mind. I still fear the future and the unknown. But in the end, who knows the future? I was 17 when my father died unexpectedly. We had everything, not a single worry in the world. Even if your life is perfect and



you're healthy, your husband is healthy, your beautiful children are healthy, it all can change in a heartbeat.

At the beginning of this diagnosis, my brother, a physician, told me that we really don't know the future of Ajda's life. He said that, in the end, knowing or not knowing one's future does not make a difference because it can all change tomorrow.

*In the end, the only constant thing in life is change.*

Everything changes – all the time. I guess the only thing that counts is what you learn along the way and how you make the best of it.

I cried when I started writing our story. But, after reaching the end, I feel strong again. We will always try to make the best of our lives. And, who knows where life will take us? I hope that my husband and I will raise Žiga and Ajda to be good, positive people so that when they are grown up, they will be able to say to themselves: "This is our life, and we choose to live it!"

**“THIS IS OUR LIFE AND WE CHOOSE TO LIVE IT!”**

# “I’m Gonna Run Right Up This Hill.”

By Stephen Jenkins, MD, Sam and Lars’ dad

SALT LAKE CITY, UTAH

The title of this article is taken from the song, “Whole Wide World,” by Mindy Gledhill, who recently did a concert to raise money for cystinosis research. You should find the song on YouTube and take a listen. It’s really good! I like the lyrics because they capture the determination, confidence and hope we need to surmount the many hills and mountains that arise because of cystinosis.

Since my son Sam was diagnosed with cystinosis in 2010, we have tried to move forward with positive attitudes and hope that he can have a happy, full life. He has overcome so much, including failure to thrive, malnutrition, feeding aversion, speech apraxia and daily episodes of nausea and vomiting. We finally reached a fairly steady state. We became used to the rigorous medication schedule, the tube feeds, the endless urinating, and the now (thankfully) less frequent episodes of emesis. When my other son Lars was born, it was a setback to learn that he, too, had cystinosis. But, fortunately, he has been blessed with good health and an iron stomach. He’s never missed a dose of cysteamine, and, to this day, his renal function is normal. He’ll probably develop Fanconi’s syndrome one day, but, if we’re lucky, he’ll never need a kidney transplant.

We were chugging along nicely, sometimes forgetting that cystinosis was part of our lives. But the last year has been filled with new challenges, and it seems that no matter how much we try to achieve normalcy, cystinosis always rears its ugly head. Sam was diagnosed with a second kidney disease called

membranous nephropathy, which results in damage to the filtering system of the kidneys and massive leaking of protein. It’s extremely rare in children, and it’s usually autoimmune or caused by a drug. In Sam’s case, the most likely cause of the membranous nephropathy is the drug cysteamine. The same drug that prolongs his life by depleting cystine from his cells is somehow triggering an immune reaction against

his kidneys. He suffered through several months of high-dose prednisone in an effort to treat it without success. It took a long time to get him off prednisone, and he had several episodes of illness requiring visits to the emergency department. He has done a couple infusions of Rituximab, and we are still waiting to see if it will work.

In all of this, we have become more committed to finding a cure. We are so grateful for the amazing work that Dr. Stéphanie Cherqui is doing at UCSD, and we are hopeful that the FDA will approve a clinical trial in 2016 for autologous hematopoietic stem cell transplant. We want to do our part to raise awareness about cystinosis and money to fund cystinosis research.

This year, we were overwhelmed by the generosity of our friends, family and strangers who came to our fundraiser. Mindy Gledhill, a popular musician in Utah, put on a benefit concert for our non-profit organization, Sam’s Hope for a Cure. My mom, Leslie Jenkins, spearheaded the event. Her cousin, Rich Winkel, is Mindy’s father. Mindy was so nice to donate all the proceeds from the concert to cystinosis research. Mindy recently bought and renovated an old blacksmith building in Provo, Utah, for her studio.



# DETERMINATION CONFIDENCE AND HOPE

CRF FAMILY STORIES



*Mindy Gledhill with  
Stephen, Lars, Ashton  
and Sam Jenkins*

She offered to perform her concert in this new space. It was the first time the venue had been used, so our event was like a special christening for the building. It was an intimate setting, and we were only able to sell 100 tickets to the show. We only advertised the event for one week before all the tickets sold out.

On the night of the event, my mom brought a small army of her close friends to help set up. They were amazing! My sister, Sarah, brought hundreds of her signature cake bites to sell, and we had dozens of cookies from Ruby Snap. We also sold Rice Krispy treats from Ashton's sister, Whitney, and cupcakes from my cousin, Emily Shaw.

We packed a little more than 100 people in the concert space. There were lots of our close friends and family members, as well as many diehard Mindy Gledhill fans. Mindy performed an amazing set, accompanied by her guitarist, Joe Corcoran. She played songs from her old albums, and the audience sang and clapped along. She dedicated a song to Samuel, and he was convinced thereafter that every song was for him. He bounced up and down in his seat, and danced with the music for the full show.

After the show, our good family friends, Rand and Lynette Patterson, presented us with a check for \$1,550 from their family business, Rand's Auto Sales. They generously pledged to donate \$25 for every car they sold in the month of September. So far, they have had a very good month!

Because we could only have 100 people at the event, we expanded our silent auction to our website. We auctioned off more than 50 auction items, including Freshly Picked moccasins, a Sonnet James dress, a Solly Baby Wrap, and multiple beautiful pieces of art. One original oil painting by David Koch sold for \$1,385!

Altogether, we raised more than \$21,000 for cystinosis research. We are so grateful to our many generous friends and family members, and in some cases, complete strangers, who have supported us in our quest to find a cure for Sam and Lars. We wish we could publicly thank every single person who helped make our fundraiser a success. Thank you to everyone who has become part of our village. You give us the hope we need to keep running up every hill and mountain that cystinosis puts in front of us.



# Hearts for Hadley

By Marcu Alexander, Hadley's mom BOISE, IDAHO

On Friday, September 25, nearly 200 guests piled into the beautiful Idaho Room of the Zions Bank Building for the 2nd Annual Hearts for Hadley Benefit in honor of Hadley Alexander. The generous crowd helped to raise more than \$68,000 for the Cystinosis Research Foundation.

The evening began with an incredible silent auction, including more than 100 unique items. Guests enjoyed appetizers and drinks while bidding on items. Five-year-old Hadley and big sister Stella were on hand to visit with old friends while also making some new ones.

The room was festively decorated with hearts and natural wood accents. Everyone in attendance received a special Hearts for Hadley wine glass to take home as a thank you for coming to support our family and cystinosis research.

Guests ate dinner while overlooking a beautiful sunset and panoramic view of Boise. After dinner, emcee and long-time friend, JJ Astorquia, introduced our family and gave Stella and Hadley a chance to thank everyone for attending before they went home with a babysitter. JJ then shared a family interview we had with news reporter Mark Johnson.

After watching the interview, JJ welcomed me back to the stage to give an update on how Hadley is doing and provide a little background on our cystinosis journey. I managed to hold it together (for the most part) and was honored to introduce my brother-in-law, Scott, to the stage to provide an update on the latest research

developments that the donations are helping to fund. Scott's speech concluded with a standing ovation and not a dry eye in the house.

Mark Johnson joined JJ on stage to help switch gears and get the party started! He played "I Gotta Feeling" by the Black Eyed Peas while getting the crowd on its feet to join him in dancing. He finished by taking off his sport coat and button-down shirt to reveal a Hearts for Hadley t-shirt underneath. After that, we had a lively dessert auction that raised more than \$3,000.

Mark & JJ both acted as auctioneers for the live auction while the guests enjoyed the treats they won. The live auction was action-packed, and the lucky winners went home with items ranging from a Seattle getaway with Seahawks tickets, to a whitewater rafting trip for two on Idaho's Middle Fork, to an event for 30 at Split Rail Winery that included appetizers and live music.

The evening concluded with a "Fund a Cure" Auction where generous supporters raised their paddles to donate directly to research for improved treatments and a cure for cystinosis.

Our family is incredibly grateful for the many people in our lives who love Hadley and who are committed to helping this cause that is so close to our hearts. In addition to the event, we received donations from people all over the country who weren't able to join us. It's overwhelming and inspiring and keeps us energized to do all that we can to help raise awareness and money.



*Our hope is abundant!*



Hearts   
for Hadley

2ND

ANNUAL  
HEARTS FOR HADLEY  
BENEFIT

1 NIGHT

25th OF SEPTEMBER

200 GUESTS

100 AUCTION ITEMS

\$68,000 RAISED

— FOR —

5 YEAR OLD HADLEY

SEE  
KEGS4KAUSE  
FUNDRAISER  
RESULTS ON  
PAGE 67!

HADLEY ALEXANDER

# Cystinosis Silver Linings

By Karen McCullagh, mom to Andrew (with cystinosis) and Kelsey, 25  
LANGDON, ALBERTA, CANADA

**W**e have all been there – hearing the horrific news that our child or loved one has just been diagnosed with a rare, life-threatening disease. We ask, “How do you spell that?” Cystinosis, cystinosis, cystinosis. We say it over and over again in our minds until eventually it becomes a part of our everyday language.

As it was for many of you, the first few months were terrifying. We reach out to grab onto anything that will help stop our world from spinning out of control.

Slowly but surely, we begin to gain ground on our new “normal.” We become more confident in our abilities to administer medications around the clock, prime overnight feeding pumps, and change urine soaked beds three times per night. We learn to juggle the demands of day-to-day life, all the while fitting in trips to the doctor, emergency room and specialists. Eventually, we take a minute, look up ever so slightly, and begin to see that, in spite of this disease, the world has not stopped turning. If we open our eyes and hearts, we find that just below the surface of this dark cloud that has hung over us for so long are small beacons of hope. I call these my “*cystinosis silver linings*.”

I didn't notice them at first, but, once I began to recognize them as the gifts they

were, I discovered silver linings all around me: my husband Don who often says, “Go to bed, babe, I'll take the night shift” even though he is dog tired from doing the early morning shift; my daughter Kelsey who is Andrew's biggest supporter and advocate; the phone calls from my mother Ann offering to help wash syringes or pull Andrew's meds; the gift of the occasional night off to spend time with my husband courtesy of my siblings Keith, Alan and Neil; Don's sisters, Joanne and Debbie, and their family who travel from BC to support us; my nieces and nephew who treat Andrew with love and normalize his childhood with rough housing, sleep overs and backyard soccer games. *Our family is a cystinosis silver lining!*

I then began to see that the community around me was rich with people who also love and support us. I am surrounded by crazy, loving, understanding women who would do anything for me. I am so blessed to call these ladies my friends. My sensei Tammy who held a kick-a-thon to raise funds for research; strangers donating money to the cause in Andrew's honor; our friends who share our story with their friends and family to help spread awareness about our little community; the players, sponsors, volunteers and donors who show up year after year to support our efforts to





Andrew, 12, and his sister Kelsey



raise funds at the JCFG Memorial Golf Tournament. There is the school staff who sit with Andrew day after day, year after year, to make sure he gets his meds at school; the educators who understand Andrew's quiet way and special learning style, and always choose "just the right teacher" for him. Our list of our community supporters goes on and on. *Our community is a cystinosis silver lining!*

I am also grateful for the opportunity to attend conferences like the Cystinosis Research Foundation's Day of Hope and meet face to face with the researchers and scientists who work tirelessly to find better treatments and the hope of a cure. They sacrifice time with their own loved ones in order to spend "just a few more minutes" for our benefit. Dr. Midgley is an angel who has been with us from the start of our journey, beginning with Andrew's diagnosis. He supports a dozen other cystinosis patients at Alberta

Children's Hospital and is always a willing listener when we share with him the "latest and greatest" news from the field. The nurses, dieticians, pharmacists, social workers, interns and other specialists support Andrew and our family daily, and surround us with information, resources and emotional support. *Our professionals are a cystinosis silver lining!*

I have been blessed with many newfound friends, and I am so grateful for the love, understanding and survival tricks I find daily in our Facebook support groups. While I am grateful to the whole cystinosis community, I hold closest to my heart the mothers of children with cystinosis – the women who walk the same path I do. I lovingly call these women my "cystas" because I know we are bonded by a shared experience that goes far beyond simply being moms. We are bound together by empathy, love and understanding. These amazing women

are my "cystahood" and united we are strong. Whenever I feel overwhelmed or down trodden, I know I can reach out and be lifted up. We can laugh, cry and know without needing to explain what "I'm tired" really means. Crystal, Kristen, Jean, Shannon, Monique, Katie, Liz, Jody, Erin, Nancy, and so many others that I have met at conferences and online, all continue to show me the strength and resiliency of a mother's love. *My cystas are a cystinosis silver lining!*

We are now over a decade into our journey. Cystinosis has become as routine to us as brushing teeth and emptying the dishwasher. We no longer live in fear and trepidation of "what might be"; but in the hope that a cure is just over the horizon. Cystinosis no longer consumes our every waking minute. Instead, we allow it only the necessary time needed to make up and administer meds, attend appointments and do what must be done to manage the disease.

There will always be days when it feels like we are surrounded by only dark clouds and gray skies. But if you open your eyes and your heart, you, too, will always find your own cystinosis silver linings.

**I LOVINGLY CALL THESE WOMEN MY "CYSTAS" BECAUSE I KNOW WE ARE BONDED BY A SHARED EXPERIENCE THAT GOES FAR BEYOND SIMPLY BEING MOMS.**

# ROCK TO THE LOCK

By Katie and Terry Monaghan, Abbi's parents

ST. CATHARINES, ONTARIO, CANADA

**W**e are Abbi's parents and, as parents, our number-one job is to take care of our kids. When Abbi is feeling well, she is a typical bubbly, energetic, nine-year-old. That said, Abbi lives every day with cystinosis.

All the families of children with this condition know cystinosis well, but we'd like to tell you more about Abbi. She is in 4th grade, and is active in gymnastics, karate, biking, swimming, crafts and music. She is a "girly-girl" who loves fashion, décor and hanging out with her friends. The burden she carries daily is the need to take upwards of 35 doses of life-saving medication in the form of capsules, liquids, pills, injections and eye drops. Much of her treatment results in unpleasant side effects.

However, Abbi's future looks brighter because of recent advancements in treatments and ongoing research, all of which bring us closer to a cure. Our positive outlook is made possible because of donations made by fundraisers.

It takes a village. In August 2015, we began planning and carrying out the early steps for an exciting, new fundraiser. The event will take place over the next 11 months and will include a series of local events to raise awareness for cystinosis and provide donations toward finding a cure.

The climax for this amazing year of fundraising will occur next summer. This is where Abbi's incredible friend Dan comes in. Dan Giancola symbolizes his motto, "Be That One." Dan was a former field goal

kicker for the Toronto Argonauts, which is part of the Canadian Football League (CFL). Today, he is an exceptional motivational and professional fitness trainer. Dan met Abbi only a year ago at a charity event, and they immediately clicked. He is Abbi's number-

one cheerleader and continues to help spread awareness, raise funds, and lift Abbi's spirits on a regular basis. The more he learns about cystinosis and Abbi's daily routine, the closer Abbi and Dan become and the more Dan wants to do for Abbi.

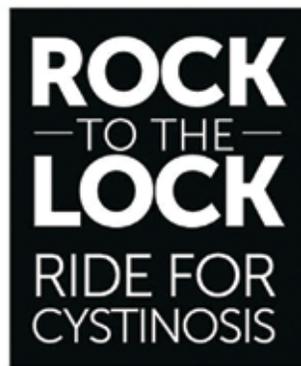
Because of Dan's commitment and relationship with Abbi, "Rock to the Lock" was born. On August 1, 2016, Dan will take on the daunting task of riding his bike from Rocks Provincial Park in New Brunswick (The Rock) to Lock 7 in Thorold, Ontario (The Lock). The cycling trek will take this self-employed husband and father 27 days. The ultimate goal for Dan is to

raise awareness and donations to help fund cystinosis research.

Our ever-growing Team Abbi is busy daily, organizing and implementing happenings from t-shirt sales, trivia nights and galas to Christmas baking and lemonade stands. We hope that a cure will come in time for Abbi to fulfill all of her hopes and dreams. We plan to be part of the cure.

*Terry and Katie Monaghan*

Follow us on Facebook, Twitter, Instagram (#Rocktothelock) and at [www.RocktotheLock.com](http://www.RocktotheLock.com)





**SEE ABBI'S  
SUNGLASS SELFIE  
FUNDRAISER  
RESULTS ON  
PAGE 69!**



**L**andon is 5 1/2 years old now and in his final year of preschool. He finished his second season of t-ball and was able to hit the ball off of the coach's pitch consistently. He just started his first season of soccer and will inform anyone who asks that he "is really good." The light in Landon shines brighter all of the time, and he makes his presence known where ever he goes. We once thought that he may be the shy child that I was, but are learning that his personality is much more outgoing, like his dad's. His health is great, and he remains, as his doctor describes, a healthy kid who takes medicine.

We have a lot to be grateful for, and I can't help but to believe that cystinosis has brought some blessings in our lives.

# LEARNING THROUGH LANDON

By Lauren Hartz, Landon's mom

PITTSBURGH, PENNSYLVANIA

In so many ways, Jimmy and I know that we are different parents than we would have been without having a child who has special needs. Landon has taught us more in his five short years than we will ever be able to teach him. For that and so much more, I am grateful.



Landon and his younger brother Jordan

*I have learned not to compare and judge milestones.*

*I have learned to function pretty well on interrupted sleep.*

*I formed a bond with my husband that I describe as a breath of fresh air. No one can comfort me quite like he can.*

*I once dreamed of being published in a magazine. I can check that one off my bucket list.*

*I have confronted my fear of public speaking. I speak about a very sensitive topic at least twice per year, and I hold myself together.*

*Our family name has been on one of those big cardboard checks when we presented the money we have raised to the Stack family at the CRF Conference.*

*I have acquired a whole new vocabulary.*

*I am stronger than I ever imagined.*

*I am an integral part of a medical treatment team, and I did not go to medical school.*

*I have met some great girlfriends who are strong and passionate, and inspire me through the cystinosis community.*

*I have spent time on the incredible beaches of California and witnessed my son say "surf's up, dudes!" to two young women with surfboards.*

*I have met young adults who are moving mountains in our world with their attitude, passion and determination. Cystinosis is just something else about them.*

*I am given the opportunity to teach people around me something new all the time.*

*Our family has experienced generosity in our community in an unbelievable way.*

*We had professionals plan our wedding of 200 people. We plan our fundraiser with twice that many people ourselves!*

*We figured out how to survive on one income, which was never our plan.*

*We are grateful for every inch Landon has grown, pound he has gained and size he has gone up.*

*We have developed a close bond with unexpected people – delivery persons, pharmacists and technicians, phlebotomists, secretaries, insurance company workers, nurses and physicians.*



SEE THE 4TH ANNUAL LOTS OF LOVE FOR LANDON GOLF OUTING RESULTS ON PAGE 67!



# JENNA & PATRICK'S FOUNDATION OF HOPE

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

## A DECADE-LONG PARTNERSHIP: JPFH AND CRF

Kevin and I have served on the Cystinosis Research Foundation Board of Directors since 2006. We recently returned from an amazing bi-annual board meeting. As we traveled home, we discussed what an honor it is to serve on the board. It is a privilege to receive regular, detailed updates regarding progress being made by those who work toward a cure for cystinosis. As board members, we are grateful to have a say in decisions that are made regarding how funds are spent, and how to best channel our energies as we work toward better treatments and a cure for cystinosis.

## THANKS TO YOU!

Since 2007, Jenna & Patrick's Foundation of Hope has raised \$1.7 million for the Cystinosis Research Foundation. Twice each year, top-tier researchers are sent official calls for research proposals, which are reviewed by the highly regarded and stringent CRF Scientific Review Board (read more about these world-renowned scientists on page 70 of this magazine). One-hundred percent of every dollar we pass along to the Cystinosis Research Foundation is used to fund studies that have the greatest potential to lead to better treatments and a cure for cystinosis and other metabolic diseases.

## IT'S GETTING REAL, PEOPLE!

We have found ourselves on the cutting-edge. Jenna and Patrick have unknowingly guided us all to become part of a ground-breaking, small-disease community. Medical discoveries made by CRF-funded scientists (and the *scientists themselves*) are being sought-after by pharmaceutical companies, larger disease communities, and the National Institutes of Health. When these entities come knocking, you

know you're on the right track! We've never had more hope, and we no longer find it unreasonable to imagine Jenna and Patrick living free from cystinosis.

## "HOW ARE THE KIDS," YOU ASK?

**Patrick** is fascinated by roller coasters and marble tracks, velocity and g-forces. He cooks a mean burger and is quite possibly the most huggable 10-year-old you'll meet. Patrick is a spiritual, mellow boy, and *he tries to negotiate everything*. He feels pretty good these days. He doesn't vomit much. He is growing and responding well to the twice-daily Procybsi® medication. It is an outrageously expensive drug (thank God for insurance), it causes a fair amount of stomach upset, and it is by no means a cure. That said, we are grateful that Patrick and Jenna both have this life-preserving medication available to them.



MARK YOUR CALENDARS FOR "SWING AND BLING #6"  
FUNDRAISER ON MARCH 10 & 11, 2016. DETAILS ON PAGE 68!



Jenna loves to move and wiggle and chat! She is a visual, tactile learner – making “sitting still and paying attention” at school a constant challenge. Her energy and happy spirit guide her as she does what she can to get the job done. Jenna is playing volleyball, enjoying the team and her time with friends. She loves fashion, make-up and baubles. She aspires to Hollywood makeup artistry... for now. Jenna is known to cook up a delightful green onion, avocado and cheddar omelet. The kids know their way around a kitchen, a skill borne of necessity, as they are both vigorous eaters.

**A HOSPITAL PEDIATRIC FLOOR IS A HUMBLING PLACE.**

A flu bug landed Jenna in the hospital for a few days in June. Hydration and electrolyte levels can be tenuous for kidney-disease patients, and too little of either can lead one down a quick, slippery slope. To spend time in the hospital is to remember Jenna's 13 days in the ICU when this journey began. We have come a long way...

**EYE DROPS EVERY HOUR?**

The cysteine-depleting eye-drops have proven to be a VERY difficult regimen. We take it a day at a time, knowing that we need to be diligent about the kids' eye care, and finding it impossible to stick to an every-hour of the day (or even a few times a day) schedule. We look forward to the nano-wafer treatment, which you can read more about on page 8 of this magazine.

Go Ghanashyam! Patrick and Jenna are ready to test out your invention!

**RESEARCH-STUDY SUBJECTS—TAKE TWO!**

The kids were invited to participate in a bone/muscle study funded by the Cystinosis Research Foundation, taking place at Stanford's Lucile Packard Children's Hospital. This relatively simple, pain-free study (that utilizes Stanford's state-of-the-art bone-scan technology) will give us a baseline of Jenna and Patrick's bone and muscle health while helping doctors better understand the implications of cystinosis on bone and muscle health. This study will ultimately encourage research into the dreaded muscle wasting that occurs in cystinosis patients' late adolescence. The kids' classmate and great pal, Nick Bruce, will jump in as a control patient for this study. *How cool is that?*

**PATRICK, JENNA, HADLEY, TINA AND HENRY IN MCCALL, IDAHO**

This summer, three other families from the cystinosis community joined us for a day on Payette Lake. We had a wonderful time celebrating summer with Hadley Alexander (p. 28), Tina Flerchinger (p. 20), and Henry (Hank) Sturgis (p. 40) and their families. The children share a special connection, and it was fun to see them together. Jenna and Patrick tried their eye drops for the first time that day in McCall, the “how to” of administering drops was lovingly demonstrated by Hadley, Tina and Hank, who helped ease

fears and cheered them on. Cystinosis has led us to some special friendships for which we are grateful.

**MAGIC-8 BALL**

Recently, Jenna and I had an outing to Target. While I was shopping, she was in the toy aisle. When I greeted her, she had been playing with the Magic-8 Ball.

Jenna was very excited to report that she'd asked it: “Will my cystinosis be cured?” and the 8-Ball answered: “There's a very good chance!”

We honestly don't talk about cystinosis much, and I was caught off guard that it was such a forward thought on her mind.

... And, we both loved the 8-Ball answer!

**TERESA TO KEVIN: “DO YOU HAVE TOPICS TO SUGGEST OR IDEAS TO EXPRESS FOR THIS CYSTINOSIS MAGAZINE UPDATE?”**

Kevin replies: “There is not an activity, an event, or a day that is untouched by cystinosis. It affects mealtime, sleep, vacations, and celebrations. Cystinosis, in some way, makes our happiest days happy, and it makes our saddest days just that. It is part of Jenna and Patrick's friendships, part of their education, and part of their extra-curricular activities. The four of us do well at pushing it to the back-burner; there is too much good in our lives to let it take us down, but there are days when it tries.”

Lovingly, and with endless thanks ...

*Teresa Partington*



WE LOOK FORWARD TO THE NANO-WAFER TREATMENT.

GO GHANASHYAM!

PATRICK AND JENNA ARE READY TO TEST OUT YOUR INVENTION!



GABBIE STRAUSS

# Running Cross Country

By Jody Strauss, Gabbie's mom  
WATERLOO, ONTARIO, CANADA

**T**his year, I signed up Gabbie and Chloe to run cross country on the KWCEA Eagles team. I had Chloe in mind when signing them up because she is naturally fast, but I didn't want to exclude Gabbie. The first practice was on one of the hottest days of summer. Deep down, I was concerned about Gabbie. Her body temperature is already warmer than normal, and now I'm going to make her run on the hottest day of the year?

We met and were welcomed by Coach Chris. He started by letting the kids know that the one thing they are not allowed to say is, "I can't." They can say, "I will try," but "can't" is not an option. I liked Chris' coaching style, and I thought we were off to a good start.

Gabbie survived the warm-ups and was holding her own, but then came time to run. A few minutes into the

running, I could see Gabbie start to slow down. Her face was beet red and kids were passing her, and I knew this was going to hurt my little competitor. You see, Gabbie doesn't play just to have fun, she plays to win. I wondered to myself, "Should she really be doing this? She has cystinosis. This was a mistake."

She came over and collapsed by my feet with barely enough energy to cry and said, "Mommy, I can't do this." I felt all the parents' eyes on me. For a moment, I thought cystinosis had won and that Gabbie's body was failing to keep up with her determined, competitive spirit. I repeated what Coach Chris said at the beginning of practice, there are no "I can't's." I told her that she can do it and that she is strong. You can try, I said. You can at least walk. My pep talk didn't seem to be working, so I

told Gabbie that she needed to make a decision to continue on the team or quit and watch Chloe every week. Not very sympathetic of me, I know.

At the end of practice, the team of tired runners gathered under a shady tree. Our experienced coach had seen these exhausted faces before and knew that he needed to deliver some inspiring words. He shared that life is a lot like running a race. Life can be hard and life can hurt, just like running, but we have to keep moving forward, no matter what. These words spoke right to my heart. Can't all cystinosis families relate? Don't our children get tired of living with cystinosis day in and day out? Don't they get tired of persistent thirst, constant medications, squinting from the sun, going to the doctor, getting poked with needles and staying in hospitals? What



about the parents and caregivers? Don't we get tired of pulling syringes, sorting pills, ordering meds, washing sheets, and visiting doctors? And what about fundraising? Yes, we are tired, but "can't" is not an option.

Coach Chris ended our time by the tree with prayer, followed by a well-deserved gummi worm for the kids. After his words of encouragement, Gabbie decided to go back and run the hills that she missed. Coach Chris told her not to look down at the rough and uneven terrain of the hill, but to keep her eyes fixed on the horizon. I believe that is what we are called to do every day. We could easily get discouraged with the challenges and daily demands of cystinosis. After all, it does feel like we are running uphill some days. But, we need to hold onto the hope that a better future is waiting for our kids. Coach Chris reminded me that we just need to look up. Beyond the hill is a beautiful horizon of hope and possibility. A cure for cystinosis is waiting; we just need to keep moving forward.

Gabbie ran some of the hill and she walked some, but, most importantly, she completed the challenge and crossed the finish line. She decided on that hot summer day that she wasn't going to give up. Since then, each practice has gotten easier, and she doesn't cry or complain anymore. I even caught a glimpse of her smiling.

I mean, after all she has endured, cross country should be no problem, right?

I love Gabbie's team t-shirt. It is her favorite color, purple, with a picture of an eagle and the words of Isaiah 40:31: *Those who hope in the Lord will renew their strength. They will soar on wings like eagles, they will run and not grow weary, they will walk and not be faint.*

Cystinosis is not for the faint of heart. Our kids are brave like eagles. They are champions and heroes. Cystinosis families are running the greatest race of their lives. With CRF leading the way, we will keep putting one foot in front of the other as we look toward the day that we cross that finish line and receive our prize – a cure for cystinosis.

**READ MORE ABOUT THE  
MOSER SUMMERFEST, HER  
FRIEND AVALEIGH, AND A  
HOMEBUILDERS SUPPORT OF**

**GABBIE'S  
WISH**

**ON PAGES 64 + 65**



24 HOURS  
FOR HANK

THE RACE ACROSS AMERICA IS OVER, BUT THE BIGGEST RACE OF MY LIFE CONTINUES UNTIL HANK IS FREE OF CYSTINOSIS.

*Hank and Grandpa, Dave Sturgis, at the Finish Line; Annapolis, Maryland, June 27, 2015.*

# THE RACE ACROSS AMERICA

# A GRANDFATHER WHOSE LOVE KNOWS NO BOUNDARIES

Photo by  
JASON DUCHOV PHOTOGRAPHY



By Dave Sturgis, Henry's grandfather  
SANDPOINT, IDAHO

**A lot of incredible stuff happened in the seven days, 17 hours and 43 minutes it took Team Laughing Dog to cycle the 3,004 miles that is the Race Across America: from a 70-year-old hitting 65 miles per hour on a descent while chasing a Brazilian rider, to the first three days of riding in temperatures where highs reached between 105 and 115 degrees, to battling torrential rains from Indiana all the way to the East Coast.**

It's tough enough to cycle across America, but Mother Nature decided to kick things up a notch. I admit, it was tough being drenched and wet for days on end, including a stretch through the rolling hills of West Virginia where I, and teammate Bob Robertson (at 71 the oldest rider in the event) climbed more than 11,000 vertical feet in 85 miles. We were pretty pleased with that accomplishment.

In addition to testing the riders, the weather wreaked havoc on the race course, forcing a couple of short stoppages and re-routes due to heavy flooding. Couple that with the intense heat early in the race, and it was easily the worst weather that The Race

Across America has experienced in its 33-year history.

One of the race's most memorable moments was when I got caught up in trying to catch the Brazilian team in front of me on a long descent, only to find out later I was clocked at 65.7 miles per hour. This septuagenarian is old enough to know better, but I got caught up in the moment. You see teams out there, you run them down, they run you down, that's part of the race's charm.

Among all of these memories, one particular moment took the cake. It came about 60 miles from Annapolis, Maryland, our final destination. A week earlier, our team, comprised of myself, Arlene Cook, Kirk Johnson, Bob Robinson and 12 crew members took off pedaling around-the-clock in six-hour shifts from the coast of California.

I was at the point of being mentally and physically drained. I was clicking off the final 100 miles in my mind, cranking on the same set of pedals my feet spent six hours a day pushing. Unknown to me, my son Brian and grandson Hank flew in and were waiting ahead at a checkpoint, a surprise visit that hit me harder than the torrential rains ever could. The emotion I experienced in seeing them hit

my heart and soul, and reminded me why I was doing something like this race.

I took on this challenge to fulfill a promise I made to Hank to find a cure for cystinosis. My biggest race still continues as we move forward to find a cure for Hank – to give Hank the opportunity to, maybe, one day race across America free of cystinosis.

As we celebrated our finish at the banquet, the race organizers called Hank up to be with the team as we accepted our finishing awards. It was pretty cool to have Hank up there with the team.

During the Race Across America, I had one goal in mind: to save my grandchild's life from a terrible disease. My promise to Hank is to make his disease go away forever. It is an investment in Hank's future and his right to a quality life.

*A grandfather is someone with silver in his hair and gold in his heart.*

–Author Unknown



7 DAYS 17 HOURS 43 MINUTES

# THE STURGIS FAMILY UPDATE

By Tricia and Brian Sturgis, Henry's parents

SANDPOINT, IDAHO

**W**e have had a great spring, summer and fall. Henry is doing well and is in 3rd grade this year. He is enjoying school and tries very hard. Henry is also participating in the after-school choir program once per week and is really enjoying all kinds of music. We continue Henry's physical and occupational therapy at school along with extra reading, math and handwriting practice in the resource room.

Henry is doing well medically. We continue to get his kidney medications compounded and made into clear, gelatin-coated pills that he can swallow easily (they used to be liquid form to take through his G-tube). He had a hard time getting the bigger pills down, especially any that have a taste. We also continue to do growth hormone shots five nights per week, which is working as he is currently in the 79th percentile for height. Henry hasn't had much of an appetite lately, so we may talk to our nephrologist about a possible medication for this.

On Father's Day weekend, Henry rode his dirt bike with Brian. It wasn't his first attempt – but definitely his longest ride so far. Dad was VERY happy!!

During our annual family camping trip to Priest Lake, Henry enjoyed playing with his cousin on the paddle boards and learned how to play tether ball. Henry tried water skiing

for the first time and did great! We are so proud of him for trying new things!

We had a very hot summer in Northern Idaho and, unfortunately, a lot of smoke from forest fires in our area. A couple of sections of nearby towns had to be evacuated. That was a reminder to be as prepared as possible in case we ever need to leave quickly in any type emergency. So, I made an emergency tote/storage box in our laundry room with some essentials for Henry. It includes a couple of doses of Henry's meds, and some water, snacks and Boost Plus, along with a doctor's note about Henry's condition. There are some quick, simple tips available at [redcross.org](http://redcross.org), if you're interested in preparing your own emergency pack.

We are looking forward to ski season this winter and skiing at Schweitzer. Our "24 Hours of Schweitzer" ski fundraiser theme this year is "Rock Around the Clock." Our goal is to have live bands, DJs or radio stations playing for the event for the full 24 hours. The fundraiser will be held on March 25 and 26, 2016.

Happy holidays and thank you to all of our friends, family and community members that tirelessly support Henry, our family and cystinosis research!

*Brian, Tricia and Henry (age 9)*



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**raptor**

# Cystinosis is Rare, and So Am I



By Shannon Paju  
ANAHEIM, CALIFORNIA

***M***y name is Shannon Paju, I am 29 years old and living with cystinosis. I reside in Anaheim, California.

I am a daughter, a cousin, and a godmother to a 15-year-old. I am a member of the Screen Actors Guild and a published poet. Cooking and making jewelry are things I enjoy doing. Because of cystinosis, I can no longer sing, dance, play musical instruments or act – all things that I love.



Shannon with her mom Bonnie

# “I MADE IT TO ADULTHOOD AGAINST ALL ODDS ... I AM WAITING TO SEE THE CURE.”



Bonnie at the Day of Hope Conference

When I was born, I was premature, and the fight for life began. At 13 months, I was rushed to the hospital where the ER doctors worked on me for nine hours before sending me to ICU. My veins had collapsed, so an IV was placed in my head. The next day, though, I was sitting in the crib like nothing was wrong! The doctors scratched their heads wondering what had just happened. The hunt was on to figure out what was wrong with me. Two weeks later, I was diagnosed with cystinosis. Yeah, me! The diagnosis was confirmed at UC San Diego. My parents were told that I had cystinosis and would most likely die by the age of three.

By the time I was 15 years old, my mom had made sure I experienced life. Starting in the sixth grade, I worked in the entertainment industry doing theatre, movies, television, and commercials. I played the flute starting in the first grade, and I also played percussion instruments.

My mom took me to Mexico, Canada, Europe and all over the United States. Life was good, then the call we had been waiting for came: we have a kidney for you. I was transplanted but, unfortunately, when I was 17 years old, the kidney died.

Cystinosis is very hard to live with. I remember the kids at school making fun of me all the time. I was made fun of because I was short, because I smelled bad from the medications, because I was allowed to leave class to go to the bathroom, or because I was allowed to wear sunglasses or to have a water bottle at my desk. There was never a day that I wasn't made fun of – not one day! Little did they know I would have loved to be normal like them.

I have had to fight for my life every day since birth. I live with pain every second of every minute, every minute of every hour, every hour of every day,

and every month of every year. There are no breaks! Between my headaches, stomach pain, back pain and depression, it's a constant battle. Just getting up in the morning is a challenge, not only for me, but for my parents who are my caregivers.

In the past 11 years, my life has been extremely hard due to my declining health. I have hemodialysis four times per week. I have been on life support 11 times. I no longer make red or white blood cells or platelets and require a blood transfusion every month. I take 66 pills each day, and 12 shots each week. In May, I was back on life support. The doctor said I would only live three days to two months. He said I should go on hospice care, but my mother worked her magic again.

Cystinosis is rare, and so am I. I made it to adulthood against all odds. I have been in clinical studies most of my life, and now we have better meds and eye drops. The meds still make me sick, but they prolong my life, and I am all about life. No two cases of cystinosis are the same, everyone is different. I am waiting to see the cure.

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TANNER EDWARDS

# A YEAR POST TRANSPLANT

By Traci Gendron, Tanner's mom  
FORT COLLINS, COLORADO

***It is now one year post kidney transplantation, and Tanner and I are doing well. We just had our one-year check-up visit to the clinic and our numbers were great! Tanner's creatinine is 1.0 and mine dropped to 1.0. The doctors said our low numbers are unheard of! It is wonderful news – we are thrilled.***

It has been a busy year! Tanner and I decided to open a menswear shop in Fort Collins, Colorado. We named the store “Storm Men’s Shop.” We chose the name because life with cystinosis can be a bit stormy at times and we thought the name had a masculine feel. There has not been a successful men’s shop in our area for quite a few years, so this is a good opportunity for us.

As we were putting the store together, we decided it would be great to give back to the cystinosis community. We decided to donate 100% of our profits to the Cystinosis Research Foundation. Our customers are thrilled with this idea! The decision to give back has given us an opportunity to talk to and educate our customers about cystinosis and our mission for a cure. They are pleased to be able to help. The store has given me and Tanner an opportunity to work together, it has given Fort Collins a much needed men’s store, and we help fund CRF research that will lead to the cure! It has been a win, win, win.

*The decision to give back has given us an opportunity to talk to and educate our customers about cystinosis and our mission for a cure.*



# *Pies and Plays for Ali and Maddie*

By Crystal and Bob Walker,  
parents to Sara (7), Aliyah (5, with cystinosis) and Madelyn (2, with cystinosis)  
CALGARY, ALBERTA, CANADA

## ***WE ALL HAVE BIG DREAMS FOR OUR CHILDREN.***

I imagine that my children will be adventurous, excited to see the world. I dream they will have compassion for others and seek out opportunities to demonstrate that compassion through action. I dream they will love learning and eagerly embrace the world they live in and the amazing things they discover. I dream that they will become fully who they are, choosing careers and life paths that give them joy and hope, that offer the best of them to the world. These dreams can become reality, with or without cystinosis.

But I also dream some things that I used to take for granted, things I couldn't imagine I would need to worry about, things that cannot be reality when living with cystinosis. I dream my children will be able to sleep

through the night most nights. I dream that their lives won't be dictated by a clock. It seems a given that our children will live longer than we do, but there it is: my biggest dream is that my children, with their adventures, courage, and joy, will outlive me, and that I will not have to live in a world without them.

Aliyah, our second daughter, was diagnosed with cystinosis in 2011, just before her first birthday. With her diagnosis, all of our dreams came crashing down around us. Suddenly, instead of dreaming and planning for the future, we were clinging just to the moment in front of us and sometimes not even that, but just to the minute in which we stood. Dreaming too far ahead was far too frightening.

I remember the day after the cystinosis test was taken, before we had results. We had been told not to research cystinosis, since it wasn't a confirmed diagnosis and we would be discouraged by all we found online. It was Saturday, and Bob was at home with Sara (our oldest, not quite 3 years old then), giving her some sense of normal after a week of living at the Alberta Children's Hospital. Exhausted from sleeping in the hospital, with its late night routines constant beeping noises, I was resting in Aliyah's room when our nephrologist came in. He smiled reassuringly, crossed his arms over the crib, and waited.

"How sure are you?" I asked him. He nodded. "Ninety-nine percent," he said. I could hardly breathe. After a few minutes, I asked another question. Dr. Wade answered, then waited for me to process and formulate another question. He stayed for what felt like hours, helping me digest all of this new information and articulate the questions that were so difficult to ask. Finally, just before he left, I asked the question that had lingered since the beginning: "How bad is this diagnosis?"

"Well," he said, with great sympathy, "there are a lot of diseases out there to have. This is a hard one. If I had to pick one, this wouldn't be it."

***I HAVE SINCE LEARNED OTHERWISE. BUT I'LL GET TO THAT.***

Those first days after diagnosis, and the first weeks and months at home, were very hard. We had shelves of medications to administer every six hours around the clock. It took four hours to wash and prepare four-days' worth of syringes that first time. We had constant appointments checking Aliyah's weight, white blood cell cysteine levels, electrolytes, growth, and

development. Aliyah threw up constantly, ate hardly anything, peed through every diaper and soaked her bed, her clothes, our clothes. Mountains of laundry. Mountains and mountains of syringes. Mountains upon mountains of questions and doubts and fears.

***TINY LITTLE MOLEHILLS OF SLEEP.***

Four years later, life is much easier, but so very different than we ever expected. Aliyah, now 5, is confident, energetic, expressive, and imaginative. She still takes mountains of syringes – 20 doses every single day. She drinks more water than your average camel. She lives with

doses. Every 6 hours, even from our sleep, our alarms remind us to give life-sustaining meds and vitamins to Aliyah and Madelyn, keeping them healthy and going strong. Even so, the disease continues to progress, causing irreversible damage that we cannot yet but will someday see.

We have met many people – over social media and in person – in our tiny cystinosis community of 2000 worldwide. We have learned the power and strength of dreaming in a different way – being part of a community that dreams with its hands. We have finally been able to join in that dreaming with

our first fundraiser – Pies and Plays for Ali and Maddie – and we watched the love and support of our friends and family grow into a beautiful reality.

*Since those first fearful days with cystinosis, we have been able to grow dreams out of hope, and are so excited to see what the future holds for all three of our girls.*

Our doctor had said that, if he had to choose, cystinosis would not be the disease

he would pick. But I have found, in our cystinosis family, a passion and call for action that I believe will lead someday to the history of cystinosis. Eleanor Roosevelt said, "The future belongs to those who believe in the beauty of their dreams." Ours is no small dream, but it is a beautiful one. And we are going to get it.

*Bob and Crystal Walker live in Calgary, Alberta, Canada with their three girls, Sara (7), Aliyah (5, with cystinosis) and Madelyn (2, with cystinosis). Pies and Plays is their first fundraiser, a family afternoon of pies, improv theatre, and silent auction. Together with the support of amazing family, friends, and "cystas," it raised just over \$26,000 for cystinosis research.*

***MY BIGGEST DREAM IS THAT MY CHILDREN, WITH THEIR ADVENTURES, COURAGE, AND JOY, WILL OUTLIVE ME, AND THAT I WILL NOT HAVE TO LIVE IN A WORLD WITHOUT THEM.***



passion and excitement, and rarely complains about meds. Two-year-old Madelyn, our youngest, diagnosed at 6 weeks, swallows her meds with a heart-breaking grimace. She has growth problems and will likely need growth hormone to help her reach her growth potential. She is a little joker, full of cuddles and love and laughs. Though these girls are little, they are fierce! As they must be.

We are very aware of the challenges – both present and future – of cystinosis. It affects every moment of every day of our lives, all five of us. Every week, we wash and fill 140 syringes.

Every second evening, we prepare 16 more. Every night, we fill another four



^ Tournament Winners – Brian DeRevere, Brad Bierbaum, Chairman Vince Ciavarella, Ryan Peterson, David Desper



^ Bob Klaewtanong, Dan Wozniak, and Tony Perez



^ Bob Cluck, John Hagestad, Tim Byrne and Joe McKay



^ Joe Oltmans, Jeff and Nancy Stack, and Randy Luce



EIGHTH ANNUAL  
**FORE a CURE**  
GOLF TOURNAMENT  
NATALIE'S WISH  
Cystinosis Research Foundation



Save the Date — Monday, October 17, 2016  
PELICAN HILL GOLF CLUB  
Contact Zoe Solsby at [zsolsby@cystinosisresearch.org](mailto:zsolsby@cystinosisresearch.org) for information



# FORE A CURE

## RAISES \$390,000

### BRINGING THE TOURNAMENT TOTAL TO MORE THAN \$2.6 MILLION

**T**he 8th Annual CRF Natalie's Wish Fore a Cure Golf Tournament held on October 19th at the Pelican Hill Golf Club was a resounding success. We had another record-breaking tournament raising over \$390,000! Led by its dynamic Chairman Vince Ciavarella, a powerhouse golf committee, an amazingly dedicated group of volunteers and the generous sponsorship support of more than 200 companies and business leaders.

"The exceptional support and generosity of our friends and community has enabled CRF to make significant advances in the treatment of cystinosis. CRF-funded researchers at institutions around the globe are making important breakthroughs toward the cure. We are honored to be a part of the progress to help Natalie's wish become a reality by contributing to cystinosis research," said Ciavarella.



After a round of golf on a beautiful day, golfers gathered for a cocktail reception and barbecue dinner outside at the golf pavilion. The festivities included a silent auction followed by a dynamic and spirited live auction. The event included many past sponsors and golfers, as well as many new supporters. The tournament's reputation as the "the best in Orange County" was confirmed as golf sponsorships were sold out several months in advance and because of our committed and energetic golf committee, most of the underwriting opportunities were sold by tournament day.

Since 2007, the Fore a Cure golf tournaments have contributed more than \$2,623,585 to cystinosis research and the quest for a cure. Our dedicated committee and volunteers, most of who have been involved since the first tournament, are the driving force behind the Fore a Cure tournament and their efforts helped make this year's event the most successful ever.



# THANK YOU TO OUR SPONSORS

## PRESENTING SPONSOR



## SILVER SPONSORS



## BRONZE SPONSORS





Tom Moran and Jason Hewitt

# THANK YOU TO OUR UNDERWRITERS

## BAG TAGS



## BEVERAGE CART HOSTS



## CIGARS



## GOLF BALLS



## GOLF CART SIGNAGE



Steve Bellitti and Tom Taylor

## GOLF CLUBS



## HATS



## LUNCH HOST



## MEDIA SPONSOR



## HOLE IN ONE



## LONGEST DRIVE



## PIN FLAGS



## SIGNAGE



## RAFFLE PRIZES



Jeff Luzar

## OWN A HOLE



## TEE SIGN



Chuck Belden





# Swing, Shoot & Liv

## GOLF CLASSIC



By Erin Little, Olivia's mom

PORT ELGIN, ONTARIO, CANADA

Once again, the small community of Saugeen Shores, Ontario Canada rallied around a little girl and her wish of curing cystinosis. The 3rd Annual Swing, Shoot & Liv Golf Classic was held on September 12, 2015 at the Saugeen Golf Club in Port Elgin with more than 175 players in attendance.

This year, the golfers were treated with blue, sunny skies followed by a BBQ lunch generously provided by The Wismer House and Hoofn' It, the local butcher. Joined by her Dada Chad, Olivia Little continued her tradition of starting the tournament with

her ceremonial tee off.

The course was filled with gimmicks, including teeing off with an Olivia-sized driver to putting with a hockey stick, as a reminder that it is a fun tournament that is all about finding a cure for a little girl.

The putting challenge consisted of a relay among each golf team. Golfers had to putt their way around the green along with enduring some of Olivia's daily challenges of what it looks like to get out the door for the day.

These activities included preparing her daily dose of medication, prepping her diaper for nap time, and filling her indispensable water bottle, then ended with dress-up time. A little girl can't get out of the house without her much

needed tutu. All of the day's activities were well received by the golfers, and they appreciated learning a little bit more about Liv and her daily challenges.

After golf, a social hour started where players and dinner guests bid on more than 100 silent-auction items, ranging from Olivia's hand-painted stools to girlfriend-getaway weekends. The auction lasted well into dinner where the winners of the day's activities were announced, including team scores and silent-auction winners. Erin and Chad Little thanked everyone, and shared Olivia's story with the crowd. Auctioneer Bill Walker concluded the evening with a couple of live auction items and Fund-A-Cure.

When the dust settled, a grand total of more than \$101,000 was raised for the Liv-A-Little Foundation. Not a bad day's work for a community of 13,000 people!

12th OF SEPTEMBER

175 PLAYERS

OVER \$101,000  
RAISED

LEARN • SHARE • COLLABORATE

CYSTINOSIS  
RESEARCH  
FOUNDATION



FIFTH INTERNATIONAL

# Cystinosis Research Symposium

BY INVITATION ONLY

SAVE THE DATES

Thursday, March 3 and Friday, March 4, 2016

We are proud to announce our 2016 keynote speaker:



**Harold M. Hoffman, MD**

Professor of Pediatrics and Medicine, University of California, San Diego

Dr. Hoffman's topic:

*Inflammasome and its association with inflammation and diseases*

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INFORMATION:

Contact Foundation Chair **Nancy Stack** at

**949-223-7610** or [nstack@cystinosisresearch.org](mailto:nstack@cystinosisresearch.org)



## CYSTINOSIS

RESEARCH FOUNDATION

RESEARCH • HOPE • CURE

[cystinosisresearch.org](http://cystinosisresearch.org)

## Volunteers Needed for a Research Study of Bone Health and Muscle Strength



Children and adults with cystinosis are at risk for musculoskeletal disease. This research study, performed by Drs. Mary Leonard and Paul Grimm, will be the first comprehensive evaluation of bone structure and muscle strength in children and adults with cystinosis. The results of this research study may be used to develop clinical trials to improve bone health, muscle strength and quality of life in children and adults with cystinosis.



### What Is the Purpose:

To examine bone structure and muscle strength in children and adults with cystinosis, compared to those without cystinosis.

### Who Is Eligible to participate:

Children and adults ages 5 to 60 years with cystinosis.

### What is required:

- Participants will be asked to complete a single study visit, lasting about 3 hours.
- Participants will be asked to complete questionnaires, physical assessments, and bone scans.
- Participants will be asked to provide a small amount of blood, about 2 table-spoons, for laboratory studies.

**For more information  
please contact:  
Jessica Whalen  
jwhalen@stanford.edu**

This investigation was supported in whole by a grant from the Cystinosis Research Foundation.

Participants will be given a picture of their bone scan, an analyzed DXA report, and \$150 gift card.

For further information regarding questions, concerns, complaints about this research or your rights, please call 1-866-680-2906



# You Can Help Find the Cure!

## 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 more than 500 registrants from 42 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.



# YOUR VOICE MATTERS

Visit the CCIR website today:

>>> [WWW.CYSTINOSISREGISTRY.ORG](http://WWW.CYSTINOSISREGISTRY.ORG)

### 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 Procybsi® 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](mailto: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?*

# Helping Us Help You!

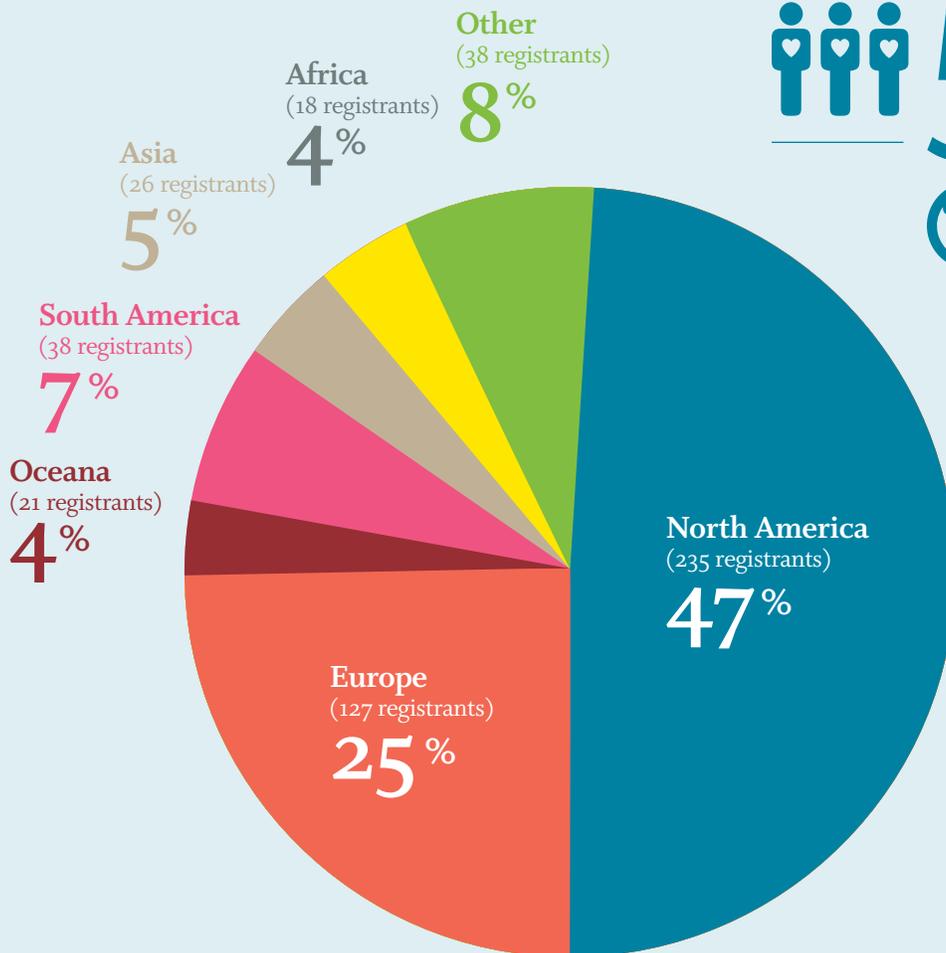
TAKING THE EXPANDED CCIR SURVEY  
BRINGS US CLOSER TO THE CURE.

REGISTER TODAY!

>>> [WWW.CYSTINOSISREGISTRY.ORG](http://WWW.CYSTINOSISREGISTRY.ORG)



## 2015 SURVEY REGISTRANTS

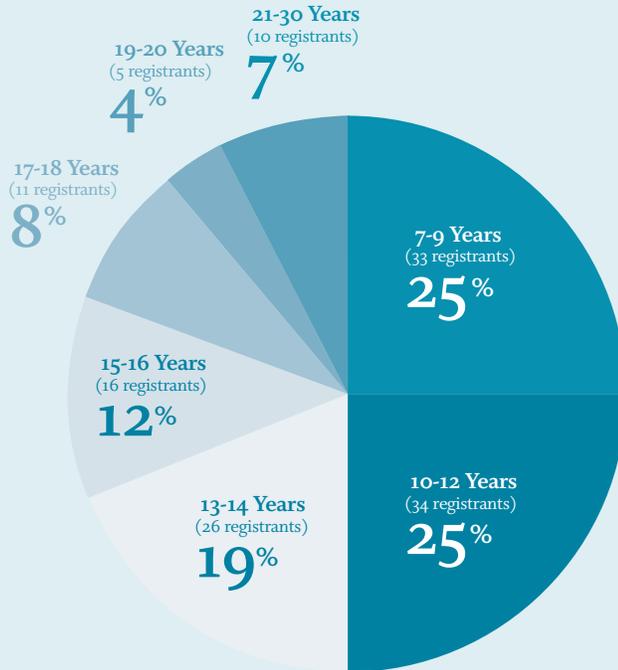


 **502**  
PEOPLE  
 from all over the world  
are part of the cure.

## KIDNEY TRANSPLANTS BY AGE

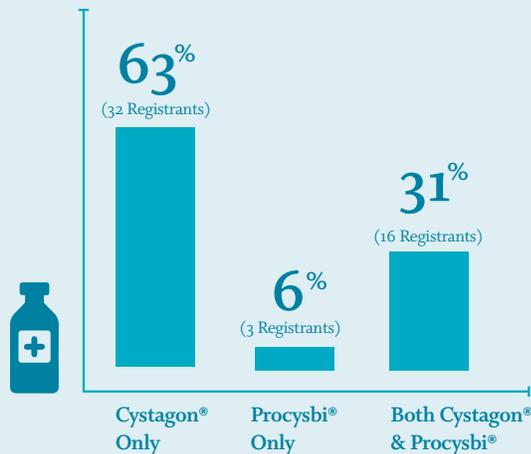


OF  
285 registrants that are  
10 years old or older,  
**153**  
have had a kidney  
transplant.  
(54%)

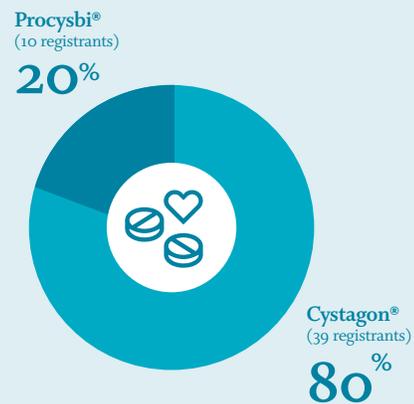


## CYSTEAMINE THERAPIES

### Experience with Cysteamine Therapies



### Current Cysteamine Therapy Taken



THESE CYSTINOSIS FRIENDS ARE

# Forever in our Hearts



## Dr. Richard Weiland

By Denice Flerchinger (Tina's mom)

I first met Dr. Richard Weiland in 2010 at our annual Tina's Hope for a Cure fundraiser. That evening, Richard and his charming wife Jenny expressed sincere concern for Tina. They told me about how inspired they felt to help us push forward toward a cure, and vowed that they would do all that they could to help. Richard and Jenny have gone above and beyond, and we could not be more grateful to have them on our side.

After meeting their grown daughter, Angie, I understood where their passion for this cause came from. Angie, like Tina, had been fighting her own battle for most of her life. When I met Angie, she had been on kidney dialysis for several years and her pancreas was failing. Thirty years of diabetes

had taken its toll on her. Richard was so proud of Angie, and he was her greatest supporter. He was by her side when she received the gift of a new kidney and pancreas last year.

Meanwhile, little did we know, Richard was fighting his own battle. On February 20, 2015, after months in and out of the hospital, Richard was taken from his earthly life. He is greatly missed.

At his family's request, memorial donations were received for cystinosis research in Richard's honor. This family has been an incredible support for many worthy causes in our local community, and we are honored to be one of them.

## Tina's Hope for a Cure



# Lynn "Pip" Ausman



When Lynn "Pip" Ausman met Tina Flerchinger at their local church, he was very touched by this special girl. Perhaps, it was because he had recently become a grandpa himself to Stone and Abby, preemie twins who were no strangers to hospitals. Maybe her medical struggles reminded him of his own daughter Lynette, who had suffered from severe asthma as a child. It might have also been Tina's shy little smile that could warm any heart. Pip understood Tina had a terrible disease, but he also knew the disease had met its match in her unstoppable spirit. He realized that if you combined that spirit with the commitment of her family and the generosity of her community, great things could happen! He became deeply engaged in the quest to find a cure for cystinosis, and was a mainstay at Tina's Hope for a Cure – a fundraising foundation established in 2009.

Pip and his wife Mary became fully engaged in Tina's mission. Mary became a member of the foundation committee

and Pip helped any way he could, including set-up, gathering of donations, clean up, and deliveries. He looked forward to Tina's auction every year, and he would be the first to get his own checkbook out, making sure that he had done his part, too. He loved that every dollar raised locally in our community went straight to the cause, and was so excited and grateful to be a part of it all. He truly believed that a cure was possible and that if he could get even one more person to participate, he could bring a cure one step closer to reality. He also thought specifically of Tina and the other affected children, and what this could mean to them. Pip loved children, and it broke his heart to hear what Tina had to go through every day and night. He remained amazed by her strength and ability to withstand what most adults could not face.

Outside of foundation work, Pip loved and enjoyed Tina and her family. They always visited at church as well as many other social gatherings. Often, they

would enjoy a special dinner together, and Pip's family would always hear about it when he "got to see Tina today." He made Tina feel very special, like he did so many children, including his own grandchildren.

Pip believed Tina would be cured, and he would be so amazed to see how close we are getting! Sadly, Pip passed away quite unexpectedly on January 20, 2015. He had an undetected case of pneumonia that had caused sepsis before it was discovered. Tina was heartbroken at the loss of this special "grandpa" in her life. The foundation chose to theme its auction in his favorite color (red) and raised a glass in his honor at the event.

One of the truest signs of a life well lived is the ability to give after you're gone. Pip was able to raise the most he ever had for Tina this year. After he passed away, family and friends donated more than \$4,000 in Pip's honor to Tina's Hope for a Cure.



## IN MEMORIAM



## CATEGORIES OF CRF RESEARCH AND NUMBER OF GRANTS

1



### CELLULAR AND/OR MOLECULAR STUDIES OF THE PATHOGENESIS OF CYSTINOSIS

30 TOTAL GRANTS

2



### STEM CELLS AND GENE THERAPY: BONE MARROW STEM CELLS, INDUCE PLURIPOTENT STEM CELLS, GENE THERAPY AND GENE EDITING

24 TOTAL GRANTS

3



### NEW DRUG DISCOVERY - CYSTEAMINE AND OTHER MEDICATIONS

20 TOTAL GRANTS

4



### KIDNEY RESEARCH

17 TOTAL GRANTS

5



### NEUROLOGICAL

11 TOTAL GRANTS

6



### CYSTINE MEASUREMENT AND CYSTEAMINE TOXICITY STUDY

8 TOTAL GRANTS

7



### EYE – CORNEAL CYSTINOSIS RESEARCH

6 TOTAL GRANTS

8



### SKIN, MUSCLE, BONE

6 TOTAL GRANTS

9



### GENETIC ANALYSES OF CYSTINOSIS

4 TOTAL GRANTS

10



### MOLECULAR STUDY OF CYSTINOSIS IN THE YEAST MODEL

2 TOTAL GRANTS

11



### THYROID

1 TOTAL GRANT

12



### CCIR

1 TOTAL GRANT

11 GRANTS

5

6 GRANTS

7

1 GRANT

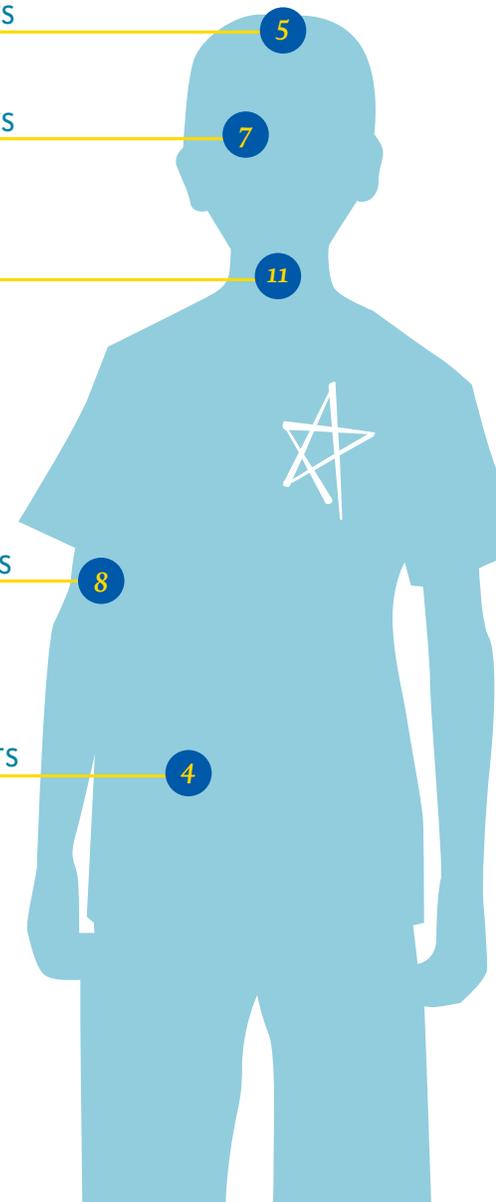
11

6 GRANTS

8

17 GRANTS

4



# CRF Research Is Providing Hope!

## 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!

CRF-FUNDED STUDIES ARE LEADING CYSTINOSIS RESEARCH

**\$29.3** MILLION IN FUNDED & COMMITTED GRANTS!

SINCE 2003, CRF HAS FUNDED **131** MULTI-YEAR STUDIES

**58** PUBLISHED STUDIES

**1** FDA-APPROVED DRUG 

**1** ALLOGENEIC STEM CELL TRIAL AT UCLA

First **CYSTINOSIS FELLOWSHIP PROGRAM**  


First **LICENSE FOR NOVEL DEVICE FOR CORNEAL CYSTINOSIS**  


**ANNUAL Day of Hope FAMILY CONFERENCE**

BIENNIAL INTERNATIONAL RESEARCH SYMPOSIUM FOR CRF-FUNDED SCIENTISTS

**1ST GLOBAL INTERNATIONAL PATIENT REGISTRY**  


ANTICIPATED IN 2016

First **AUTOLOGOUS STEM CELL AND GENE THERAPY TRIAL**

# TOGETHER, WE ARE **One**



GABBIE STRAUSS



## MOSER SUMMERFEST IN SUPPORT OF GABBIE'S WISH



Once again, the Moser family has come to Gabbie's side by hosting another fundraiser in support of cystinosis research. Summerfest weekend was held August 1 and 2, 2015. Friends and family of the Moser's enjoyed camping, beef BBQ, live music, swimming and fun for a great cause. **More than \$1,300 was raised.** We are so thankful to Melanie and Jeff Moser for their steadfast support over the years.

## AN AMAZING RACE BIRTHDAY WISH

IN HONOUR OF  
GABBIE STRAUSS



Gabbie's friend, Avaleigh Hansen, attended the **Wizard of Oz** fundraiser for cystinosis in March. After hearing more about the disease, Avaleigh turned to her mom and said, "I didn't know Gabbie was sick." Despite being only 7 years old, Avaleigh wanted to do her part to help. In lieu of birthday gifts for her 8th birthday, Avaleigh requested donations to Gabbie's Wish. The theme of her party was The Amazing Race. Amazing is a great word to describe Avaleigh and her entire family. We have been so blessed by their support and prayers. The Hansens are one AMAZING family.

# 1 PURPOSE. 1 JOURNEY. 1 CURE.

GABBIE'S WISH

## HOMEBUILDERS CARE FOR GABBIE'S WISH



On September 22, The Waterloo Region Home Builders' Association (WRHBA) held its annual Charity Golf Classic at the beautiful Whistle Bear Club in Cambridge, Ontario. The Association's Executive Director Marie Schroeder opened her heart to our story and family. Gabbie and Chloe dressed up and enjoyed the beauty of the venue, the attention from all the wonderful golfers and playing with Marie.



The event raised \$5,000 for cystinosis research.

WRHBA's President, Graem Jackson presented a \$5,000 check to the Strauss Family in recognition of Gabbie's Wish.

MORGAN PEACHMAN

## MULLIGANS FORE MORGAN



We held our first annual Mulligans Fore Morgan golf outing, in honor of our daughter Morgan Peachman, on Sunday, September 20, 2015, at Bob-O-Link Golf Course in Avon, Ohio. With 88 golfers, 26 hole sponsors, and many friends and family members volunteering their time to make the event a success, we were able to **raise more than \$12,000 for the Cystinosis Research Foundation.**

Morgan started the golf outing, hitting her ceremonial tee shot while our field of golfers watched and cheered. She hit a pink golf ball and made great contact on her drive, thanks to her golf coach, her Grandpa Scott Saari. "Team Morgan" wore pink Mulligans Fore Morgan shirts to show our support for Morgan.

Morgan had a wonderful time working at her lemonade stand and giving out "medicine" to the golfers on the 8th hole. Her favorite part of the event was watching the golfers take their medicine when she told them to, talking on the microphone during the raffle and thanking everyone for coming out to the event.

On behalf of Morgan, the Peachman and Saari families, and the Cystinosis Research Foundation, thank you to all who participated in and supported our event. We can't wait to see you at the 2nd Annual Mulligans Fore Morgan golf outing in 2016!



# TOGETHER, WE ARE One

IN HONOR OF JAKE KRAHE

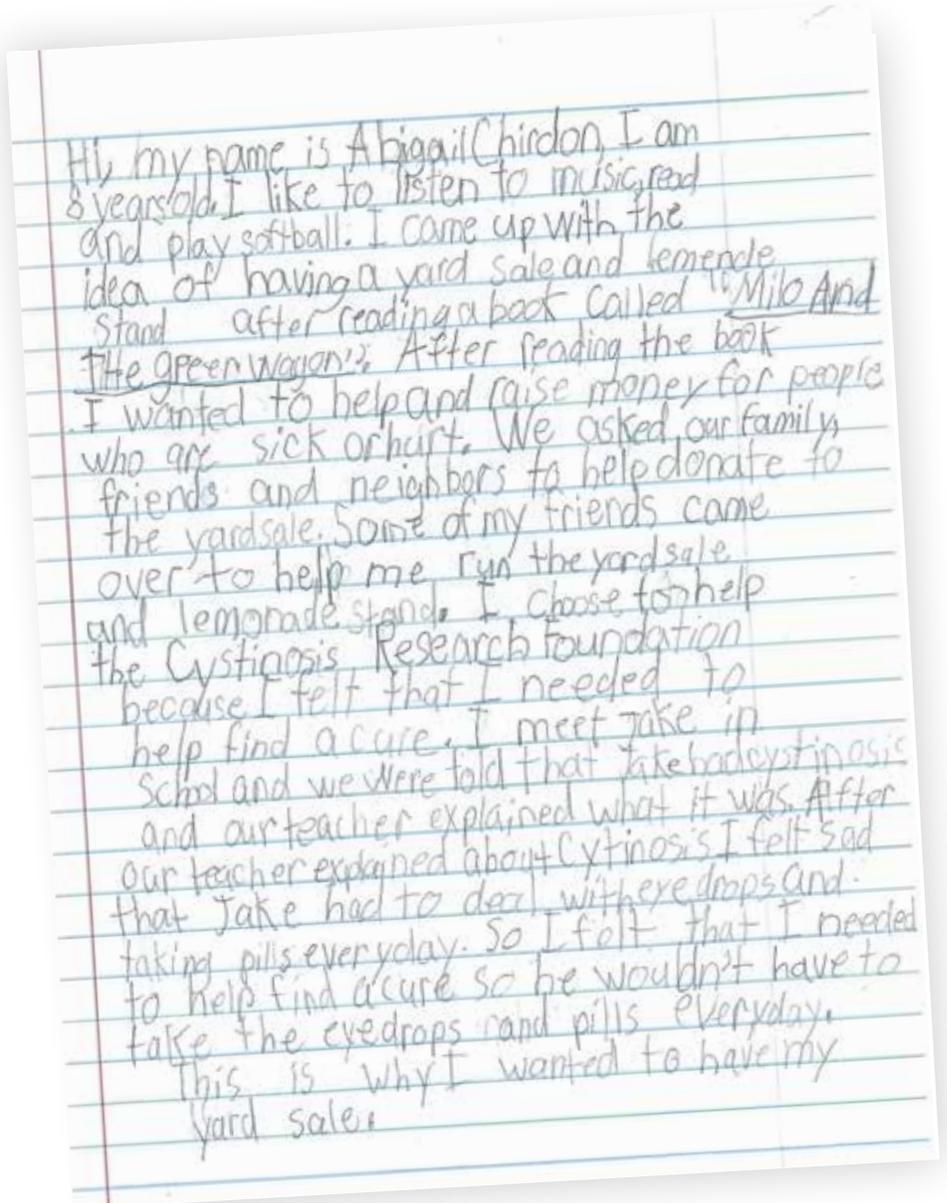
ABIGAIL CHIRDON'S

## BIG HEART

Abigail Chirdon is a very special little girl with a very big heart. She came up with the idea to conduct a yard sale and lemonade stand to raise money for the Akron Children's Hospital activity fund.

The fund enables the hospital to purchase toys and activities to help keep young patients occupied during their treatments. In addition, during the last two years, Abigail has done a toy-collection drive among her family and friends at Christmas time where she donated the gathered items to the hospital.

This year, Abigail did the yard sale and donated the proceeds to the Cystinosis Research Foundation to help her friend Jake Krahe, who has the disease. She looks forward to doing the yard sale and lemonade stand in future years to benefit worthy causes, such as the good work of the Foundation.



# 1 PURPOSE. 1 JOURNEY. 1 CURE.

HADLEY ALEXANDER

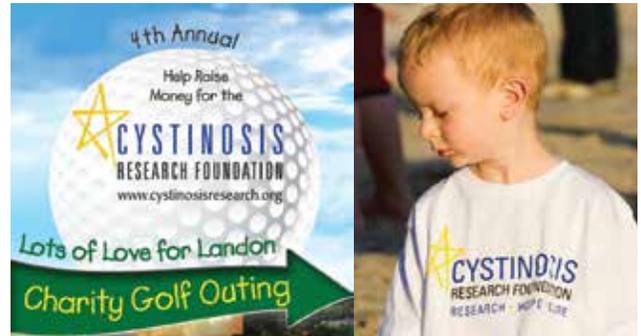


Over the past few years, we've learned that there are many interesting ways to raise money for cystinosis. Supporters have arranged poker tournaments, lemonade stands, music concerts and volleyball tournaments, among other events, to raise funds. Thanks to Hearts for Hadley, we can now add beer drinking to that list!

On Monday, August 3, 2015, Payette Brewing Co. in Boise, Idaho, welcomed Hearts for Hadley as its selected non-profit for Kegs4Kause. Kegs4Kause is a unique, recurring event created by Payette Brewing as a way to give back to the community. Every Monday evening, 50 percent of all proceeds from beer sales are donated to a featured non-profit organization.

We were honored to be selected by Payette Brewing and enjoyed a fun evening surrounded by friends and family who came to support Hearts for Hadley while drinking some delicious craft beer. There was also a food truck onsite for guests to enjoy dinner. As a result of the event, **Hearts for Hadley raised \$1,837** from beer sales, Hearts for Hadley t-shirts sales, and generous donations. *We love our supportive community!*

LONDON HARTZ



Family and friends of Landon Hartz are proud to have sponsored the 4th Annual Lots of Love for Landon Golf Outing. The new venue was at Blackhawk Golf Course in Blackhawk, Pennsylvania, and more than 100 golfers attended. Golfers had many chances to go home with a prize between 50/50 raffle, Chinese Auction and door prizes. Jimmy, Lauren, Landon and Jordan Hartz are thankful for the hard work of Jason Hartz, Jason Whitfield and Brad Hamilton in planning this event. In total, **\$20,000 was raised and donated to the Cystinosis Research Foundation.**



Jason Whitfield, Jason Hartz, and Brad Hamilton with Landon!

# TOGETHER, WE ARE One

✓ **MARK YOUR CALENDARS!**

MARCH 10 & 11, 2016



“Swing”

Golf at Catta Verdara on Thursday,  
March 10, 2016



“Bling”

Dinner and Auction at The Citizen Hotel  
on Friday, March 11, 2016



100% of funds  
raised will be passed onto the  
Cystinosis Research Foundation



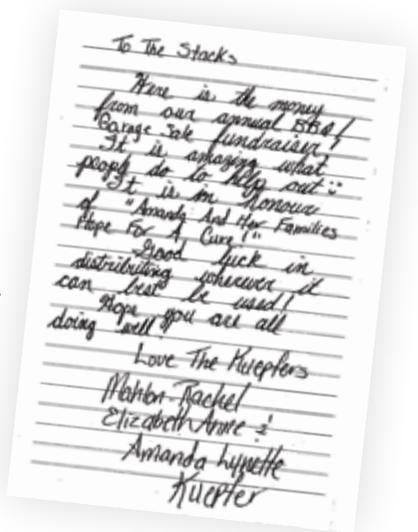
AMANDA KUEPFER



## BBQ+GARAGE SALE

The Kuepfer Family of Ontario, Canada, hosted its annual BBQ and garage sale this spring in honor of their daughter Amanda. The Kuepfer's family and friends celebrated Amanda and her family's Hope for a Cure by showing their support and generosity, raising **\$4,655 for cystinosis research.**

It is amazing what a small group of committed individuals can accomplish! On behalf of the CRF and the cystinosis community, thank you.



KUEPFER FAMILY  
ONTARIO, CANADA

08 JULY 2015

PAID TO THE Cystinosis Research Foundation \$ 4,655.00  
CHECK OF - four thousand six hundred fifty five - 00 DOLLARS @

for the cure!

With Love

# 1 PURPOSE. 1 JOURNEY. 1 CURE.

ABBI MONAGHAN



## SHOW US YOUR SUNGLASS SELFIES!

For the past three years, Abbi's school has supported spreading awareness and raising funds for cystinosis by hosting a "Sunglass Day" at school on Abbi's birthday. Children donate a couple of dollars to wear their shades to school for the day. This past year, we decided to step it up and use social media to help spread awareness. We launched a Facebook event, sent out invites and, within a week, had more than 1,600 participants with 14,000 invites!

We asked people to take a selfie while wearing sunglasses, post it to Twitter, Instagram or Facebook with the hashtag #CystinosisSunglassDay2015 and donate \$5 to the Cystinosis Research Foundation in Abbi's honor. We had pictures of animals, babies, friends, family, and complete strangers. They all had a great time, as they helped spread awareness. Our hope was to get Taylor Swift to wear some shades and snap a selfie. It didn't happen this year, but there's always Abbi's next birthday. We won't stop working to raise awareness until a cure is found!

## #CYSTINOSISSTRONG

BY JESSICA JONDLE

During the month of September, there was a #CystinosisStrong campaign on social media aimed at raising awareness of how we can combat muscle wasting. The campaign asked people to snap a photo of them doing something strength building, or of something that symbolized strength to them, and tag it with the hashtag #CystinosisStrong. Participants were also asked to donate \$10 to the Cystinosis Research Foundation. Photos that included a person with cystinosis doing something strength-building gained an extra entry into the campaign's "photo of the week" competition!

Muscle wasting is a serious concern for those living with cystinosis beyond childhood. The community has long been in need of more research into this area, and it is encouraging that the Cystinosis Research Foundation is funding another study related to muscle wasting. The CRF has previously been an advocate in this area, with Dr. Trauner laying the groundwork for future studies. We are so grateful to CRF for demonstrating its commitment to both children and adults who live with cystinosis.

The purpose of #CystinosisStrong was to give back to CRF in a show of support for this important research and raise awareness of an issue that is often in the background due to the more urgent post-diagnosis concerns of electrolyte management and kidney maintenance. As a community, we will work together to find treatments that lead to a better quality of life for those of all ages.



## SUNGLASS SELFIES

## LEADERSHIP. GUIDANCE. COMMITMENT.

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



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*Professor*

*Laboratory of Hereditary Kidney Diseases  
Imagine Institute (Inserm U1163)  
Paris, France*

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*Professor of Pediatrics*

*Ohio State University  
Columbus, Ohio*

*Thank you for your dedication to  
the global cystinosis community.*

TOTAL: **\$1,405,420**

## 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)

## 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)

## 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)

## 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)

## 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)

## 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, 2016 – January 30, 2018)

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

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

## DEVELOPMENT OF TRANSDERMAL CYSTEAMINE DELIVERY SYSTEM

Ghanashyam Acharya, PhD, *Principal Investigator*

BAYLOR COLLEGE OF MEDICINE, HOUSTON, TEXAS

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### OBJECTIVE/RATIONALE:

The objective of this project is to develop a transdermal cysteamine delivery system (TCDS) to treat nephropathic cystinosis. Currently available cysteamine oral pills cause side effects such as vomiting, abdominal pain, diarrhea, loss of appetite, and breath and skin odor. To circumvent these drawbacks, a transdermal delivery system will be developed: the TCDS containing cysteamine will be affixed to the skin and cysteamine will be released for up to 24 hours. The new TCDS will enhance drug efficacy and treatment compliance.

### PROJECT DESCRIPTION:

The two specific aims for this project are:

(1) Fabrication and characterization of TCDS

The TCDS will be fabricated with various amounts of the drug and polymer matrices to find a composition that allows adequate dose of cysteamine to diffuse through the skin. The fabricated TCDS will be thoroughly characterized to ensure the final content of cysteamine and its long-term stability.

(2) Optimization of *in vitro* drug release kinetics

*In vitro* cysteamine release kinetics from the TCDS will be evaluated using Franz diffusion cells. Briefly, the TCDS will be affixed on a silicone membrane or human cadaver skin, after which the diffused cysteamine will be collected and quantified. Hence, cysteamine release and diffusion rate can be optimized further.

### RELEVANCE TO THE TREATMENT OF CYSTINOSIS:

The TCDS will deliver cysteamine via skin in a more effective and efficient way, while minimizing the undesirable side effects. Upon successful development, the TCDS will be a new treatment modality for nephropathic cystinosis. As an alternative to commercially available oral cysteamine pills, the TCDS offers many advantages; the TCDS as a patch is non-invasive, disposable, inexpensive, and self-administered, thus improving the quality of life for cystinosis patients.

### ANTICIPATED OUTCOME:

The developed TCDS will release cysteamine for up to 24 hours via skin that can maintain a constant cysteamine concentration in the blood stream. Successful completion of this project will provide valuable information that enables the fabrication of the TCDS for *in vivo* animal studies and human clinical studies in the future.

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

Stéphanie Cherqui, PhD, *Principal Investigator*  
UNIVERSITY OF CALIFORNIA, SAN DIEGO

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### OBJECTIVE/RATIONALE:

The extent of efficacy of hematopoietic stem cells (HSCs) to rescue cystinosis was surprising, especially considering that cystinosis is a transmembrane lysosomal protein expressed in every tissue. The objective of this project is to investigate the mechanism by which HSC transplantation can lead to long-term tissue repair in cystinosis.

### PROJECT DESCRIPTION:

Addressing the mechanism of tissue repair by HSCs in the mouse model of cystinosis, we recently showed that HSCs differentiate into macrophages that generate long, tubular protrusions called tunneling nanotubes (TNTs) that mediate the transfer of “healthy” lysosomes to the adjacent disease cells. Macrophages, the most plastic cells of the hematopoietic system, are found in all tissues and show great functional diversity. In addition to the maintenance of homeostasis and immune function, macrophages have emerged as important therapeutic targets in many human diseases. Therefore, we propose to determine the phenotype of the macrophages capable of generating TNTs and involved in the tissue preservation. We also propose to identify key proteins involved in the formation of these TNTs. Finally, we will investigate the mechanism involved in HSC-mediated glomerular rescue in the mouse model of cystinosis.

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

This work will enable the understanding of the mechanism by which HSC transplantation rescues cystinosis. This knowledge will be important for the future stem-cell gene therapy clinical trial for cystinosis. This work will open new perspectives in regenerative medicine that could spur the development of novel stem cell-based therapies for other lysosomal disorders.

### ANTICIPATED OUTCOME:

We expect to identify what subtype of macrophages is responsible for tissue repair in cystinosis and what key proteins are involved. We will also determine how glomeruli, important structures in the kidney, are preserved after HSC transplantation.

## MOLECULAR MECHANISM OF CYSTINOSIS

Liang Feng, PhD, *Mentor*

Xue Guo, PhD, *Research Fellow*

STANFORD UNIVERSITY

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### OBJECTIVE/RATIONALE:

Cystinosis is caused by abnormal buildup of cystine inside the lysosome. Understanding how a small molecule like cystine is transported across the membrane is critical to understand the cause of and potential therapies for cystinosis. Our goal is to understand the way small molecules are transported across lysosomal membranes at the molecular level using multidisciplinary approaches. Specifically, we are going to “visualize” transporter proteins in atomic detail to find out how they carry out their function.

### PROJECT DESCRIPTION:

Our goal is to provide a blueprint of the transport protein and to figure out how the substrate is transported. We will first produce large amounts of the transporter protein in cells and separate them from impurities by protein purification. Then we will identify conditions that enable the protein to form ordered 3D crystals. After that, we will analyze the protein crystals using a bright and focused x-ray beam in order to deduce structural information and build an atomic model for the transporter protein. In the meantime, we will establish a method to measure the transport activity in a well-defined system. We will also pinpoint components of the protein that are critical for transport activities.

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

Despite past efforts on functional studies, many critical aspects about the membrane transport process relevant to cystinosis remain unknown. Our research will address two fundamental questions: how small molecules are brought into and out of lysosomes and how we can manipulate the membrane transport process for cystinosis treatment. The knowledge learned here will provide insight into the transport mechanism and aid in the discovery of novel therapeutic agents.

### ANTICIPATED OUTCOME:

In this study, we expect to decipher the molecular basis of membrane transporters by obtaining their atomic models. What’s more, through structure-guided functional analysis, we will be able to identify molecular determinants of transport function. Such information will reveal molecular properties of lysosomal membrane transporters and help us to better understand both the cause and treatment of cystinosis.

## NEWBORN SCREENING FOR CYSTINOSIS

**Sihoun Hahn, MD, PhD, *Principal Investigator***  
SEATTLE CHILDREN'S HOSPITAL

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### OBJECTIVE/RATIONALE:

Despite the fact that cystinosis can be effectively treated with an excellent outcome if diagnosed early, it is unfortunate that there are currently no cost-effective screening methods available for early detection to prevent from developing permanent complications such as kidney failure. This project is intended to develop a high-throughput and multiplexed assay using tandem mass spectrometry to quickly screen cystinosis using dried blood spots for potential newborn screening.

### PROJECT DESCRIPTION:

Most mutations causing cystinosis in *CTNS* gene result in absent or reduced CTNS protein; thus CTNS protein itself has an enormous potential as a biomarker for the screening of cystinosis. In addition, the 57-kb deletion, the most common mutation includes the entire flanking *SHPK* gene, which encodes the enzyme sedoptulokinase (SHPK). Therefore, SHPK can be an additional valuable marker for patients with homozygous 57-kb deletion. Our goal is to develop an immuno-SRM assay to identify and quantify a panel of signature peptides for CTNS and SHPK in DBS and facilitate the early detection of cystinosis in newborn period. The SRM-MS assay, by coupling with peptide immunoaffinity enrichment (immuno-SRM) for CTNS and SHPK, will be tested for the ability to correctly identify the patients with cystinosis.

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

Our proposal to develop an immuno-SRM assay to identify and quantify a panel of signature peptides for CTNS and SHPK in DBS would create an innovative tool to screen cystinosis in newborns. Early detection of cystinosis soon after birth, before developing any severe complications, would offer the best chance for successful and effective treatment. Moreover, accurate quantification of CTNS can be utilized as a powerful tool to monitor the drug efficacy for potential new therapy such as chemical chaperons.

### ANTICIPATED OUTCOME:

With respect to **expected outcomes**, the established quantitative immuno-SRM-MS assay will have the potential to increase our capacity for early detection of cystinosis. Such results will provide a testing methodology that is viable both for NBS and for rapid diagnosis for older individuals. In addition, this research may be applicable to many other congenital disorders providing the opportunities for early intervention thereby increasing survival and decreasing morbidity.

## A STUDY ON BONE REMODELING DEFECTS IN NEPHROPATHIC CYSTINOSIS

**Anna Taranta, PhD and Andrea Del Fattore, PhD,**  
*Principal Investigators*

BAMBINO GESÙ CHILDREN'S HOSPITAL, ROME, ITALY

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### **OBJECTIVE/RATIONALE:**

Hypophosphatemic rickets (HR) is frequently observed in patients with nephropathic cystinosis (NC). HR is characterized by impaired mineralization of bone matrix that leads to skeletal abnormalities. To date, HR in patients with NC has been thought to be mainly caused by renal tubule damage and consequent loss of phosphate in the urine. However, rickets described in cystinotic mice was not associated with renal dysfunction. The aim of this study is to identify other factors that contribute to bone remodeling defects in NC.

### **PROJECT DESCRIPTION:**

This project proposes to study bone remodeling defects in male *Ctns*<sup>-/-</sup> mice that do not manifest renal dysfunction before nine months of age. In particular, the first aim is to describe the bone phenotype and to evaluate the production of systemic hormones and paracrine/autocrine factors which regulate bone remodeling in cystinotic mice at different ages. The second aim is to study *in vitro* the role of cystinosin on differentiation and activity of bone cells. To this purpose, we will analyze the expression of transcriptional factors that regulate the differentiation, the expression of bone matrix proteins involved in the mineralization, the production of factors that control osteoclastogenesis, resorption efficiency of osteoclasts in the mineralized and demineralized bone, and the regulation of secretory pathways which are extremely active in bone cells.

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

This study on bone remodeling defects in NC will allow us to understand how the cystinosin deficiency influences the processes of differentiation, formation/mineralization and resorption of bone cells. The identification of factors that are responsible for bone remodeling defects in NC will provide useful information on potential targets for new therapeutic approaches. The final purpose of this project is to find ways to improve the bone health and consequently the quality of life of the patients with NC.

### **ANTICIPATED OUTCOME:**

Reproducing a bone *in vitro* model, without the influence of signals induced by renal dysfunction, we expect to discover how the cystinosin deficiency can modify the activities of formation/mineralization and resorption of bone cells. Moreover, by *in vivo* study on cystinotic mice, we expect to know if the defective expression of bone-secreted factors can contribute in developing extra-renal complications as diabetes mellitus and male infertility. This study will allow us to better understand the causes of HR in the NC.

## LEPTIN SIGNALING IN INFANTILE NEPHROPATHIC CYSTINOSIS (INC)

**Robert Mak, MD, PhD, *Principal Investigator***  
UNIVERSITY OF CALIFORNIA, SAN DIEGO

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### OBJECTIVE/RATIONALE:

Inflammation has been implicated in muscle wasting and kidney disease progression in infantile nephropathic cystinosis (INC). Leptin regulates energy homeostasis and is a key immunomodulatory cytokine. Our preliminary results in *Ctns*<sup>-/-</sup> mice, an established murine model of INC, suggest that leptin signaling is important for INC-associated cachexia. We propose to test whether pharmacologic and genetic blockade of leptin signaling may ameliorate muscle wasting and kidney disease severity in INC. The results of this investigation may pave the path for novel therapies for INC.

### PROJECT DESCRIPTION:

We propose to investigate whether treatment with a leptin receptor antagonist could ameliorate INC-associated cachexia. Leptin exerts its hypothalamic effects through melanocortin type 4 receptor (MC4R). AgRP is an antagonist of MC4R. The effect of melanocortin receptor blockade in INC-associated cachexia will be tested using both genetic (by generating *Ctns*<sup>-/-</sup>MC4R<sup>-/-</sup> mice) and pharmacological approach (by administration of AgRP to *Ctns*<sup>-/-</sup> mice). Leptin has nephrotoxic effects. We will test whether pharmacologic and genetic blockade of leptin signaling may ameliorate kidney disease severity and progression in *Ctns*<sup>-/-</sup> mice.

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

Muscle wasting is a common complication in patients with INC. Muscle wasting, weakness and the consequent loss of mobility negatively affect the quality of life. The underlying mechanisms of muscle wasting in patients with INC are not well understood. Leptin signaling may be important for the pathogenesis and complications of INC. This project is important as we investigate the role of leptin and MC4R signaling in INC. Our hypothesis, if proven, will likely lead to novel therapy for INC.

### ANTICIPATED OUTCOME:

Using both genetic and pharmacological approach, we intend to block leptin signaling in mouse model of INC. We expect to observe a significant improvement in metabolic and renal phenotype in cystinosis mouse. Results of this proposal may pave the way for novel therapy for INC-associated cachexia and progression of kidney disease for which there are no targeted treatments.

ACTIVITIES



CALENDAR

Thursday, March 3 and Friday, March 4

**CRF INTERNATIONAL  
CYSTINOSIS RESEARCH  
SYMPOSIUM** *for CRF-funded  
Researchers and Scientists*

Arnold and Mabel Beckman Center  
Irvine, California



Thursday, March 10 and Friday, March 11

**SWING AND BLING FUNDRAISER  
FOR JENNA AND PATRICK PARTINGTON**

Golf at Catta Verdara / Dinner Gala at  
Citizen Hotel, Sacramento, California

[www.jpfh.org](http://www.jpfh.org)



MAR  
2016

March 2016

**EASTER EGG-STRAVAGANZA  
FOR OLIVIA LITTLE**

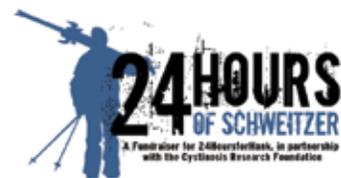
Liv-A-Little Foundation  
[www.livalittlefoundation.com](http://www.livalittlefoundation.com)



Friday, March 25 — Saturday, March 26

**24 HOURS OF  
SCHWEITZER SKI EVENT,  
ROCK AROUND THE CLOCK  
FOR HENRY STURGIS**

Schweitzer Mountain, Sand Point, Idaho  
[www.24hoursforhank.org/events](http://www.24hoursforhank.org/events)



Thursday, April 7 — Saturday, April 9

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## CRF DAY OF HOPE FAMILY CONFERENCE

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The Island Hotel, Newport Beach, California

For more information, contact Nancy Stack, [nstack@cystinosisresearch.org](mailto:nstack@cystinosisresearch.org)



Saturday, April 9

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## CRF NATALIE'S WISH CELEBRATION

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The Island Hotel, Newport Beach, California

For information or sponsorship information, contact  
Zoe Solsby, [zsolsby@cystinosisresearch.org](mailto:zsolsby@cystinosisresearch.org)



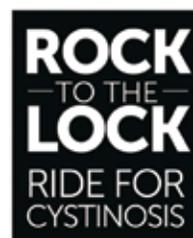
Monday, August 1 — Saturday, August 27

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## ROCK TO THE LOCK FOR ABBI MONAGHAN

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Bike ride from Provincial Park, New Brunswick  
to Lock 7 in Thorold, Canada [www.rocktothelock.com](http://www.rocktothelock.com)



Saturday, September 10

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## SWING, SHOOT & LIV GOLF CLASSIC FOR OLIVIA LITTLE

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Liv-A-Little Golf Tournament, Port Elgin, Ontario, Canada  
[www.livalittlefoundation.com](http://www.livalittlefoundation.com)



September

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## FOREFATHERS GOLF TOURNAMENT FOR ANDREW CUNNINGHAM

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Boulder Creek Golf Course, Langdon, Alberta, Canada  
For information email Karen McCullagh: [kcm\\_consulting@msn.com](mailto:kcm_consulting@msn.com)



[www.facebook.com/JCFGMemorialgolf](http://www.facebook.com/JCFGMemorialgolf)

Monday, October 17

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

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Pelican Hill Golf Course, Newport Beach, California



## CRF Is Eager 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 \$29 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 March 2016, CRF will announce its spring 2016 call for research proposals and fellowships. Details and guidelines for applications will be available online at the CRF website:

[www.cystinosisresearch.org/research/for-researchers](http://www.cystinosisresearch.org/research/for-researchers)

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.

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 more than 500 registrants from 42 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](http://www.cystinosisregistry.org).

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.

### PUBLISHED STUDIES BY CRF-FUNDED RESEARCHERS

CRF-funded researchers have published 58 studies, including these since the Spring 2015 issue of Cystinosis Magazine!

#### Treatment of Inherited Eye Defects by Systemic Hematopoietic Stem Cell Transplantation

Published in *IOVS, Investigative Ophthalmology & Visual Science*  
by Stéphanie Cherqui, PhD  
UNIVERSITY OF CALIFORNIA, SAN DIEGO



#### Time Course of Pathogenic and Adaptation Mechanisms in Cystinotic Mouse Kidneys

Published in *JASN, Journal of the American Society of Nephrology*  
by Héloïse Chevronnay, PhD, and Pierre Courtoy, MD, PhD  
DE DUVE INSTITUTE, BRUSSELS, BELGIUM



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MISSION

The mission of the Cystinosis Research Foundation is to find better treatments and a cure for cystinosis by supporting bench, clinical and translational research. Since 2003, CRF has raised \$29.3 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.

SAVE THE DATE

*The year's most inspiring and exciting celebration!*

2016  
**NATALIE'S WISH  
CELEBRATION**

FEATURING  
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**RACHEL PLATTEN**

BLOCKBUSTER HIT  
"FIGHT SONG"

NEW RELEASE  
"STAND BY YOU"

SATURDAY  
APRIL 9, 2016

THE ISLAND HOTEL

NEWPORT BEACH  
CALIFORNIA

For sponsorship opportunities or tickets, contact Zoe Solsby at 949 223 7610 or [zsolsby@cystinosisresearch.org](mailto:zsolsby@cystinosisresearch.org)