Cystinosis MAGAZINE

SPRING 2015 FOR FRIENDS AND SUPPORTERS OF THE CYSTINOSIS RESEARCH FOUNDATION

Currently, there is no cure for cystinosis but there is

Hope

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

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

Cystinosis affects approximately 500 people, mostly children, in North America and fewer than 2,000 worldwide. It is one of the 7,000 rare or "orphan" diseases in the United States that collectively impacts approximately 30 million Americans.

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

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

Today, CRF is the largest provider of grants for cystinosis research in the world, funding more than 125 studies in 12 countries. CRF has raised $29.3 million, which it has granted or committed to cystinosis research studies around the world. CRF's efforts have changed the course of cystinosis research and given new energy to its investigators and scientists. CRF's commitment to research has given hope and promise to the global community of cystinosis patients and their families.

CYSTINOSIS
RESEARCH FOUNDATION
RESEARCH • HOPE • CURE

cystinosisresearch.org

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On the cover:
Jenna and Patrick Partington enjoy playing after a spring time shower. The beautiful photo was taken by April Melarkey, a longtime supporter of Jenna & Patrick's Foundation of Hope.

Please send your suggestions and comments regarding *Cystinosis Magazine* to nstack@cystinosisresearch.org.

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

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

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Dear Friends and Family:

EMBRACED BY OUR FAMILY AND FRIENDS

The gorgeous and playful picture of 10-year-old twins, Jenna and Patrick Partington, light up the cover of this issue of *Cystinosis Magazine*. Their mother, Teresa Partington, shares a poignant letter written to the twins about the change in their current medical treatment and her thoughts on their future (page 34).

As you read through the articles in this issue of the magazine, you might notice a common thread: the importance of our family and friends, who are integral parts of our journey with cystinosis. As a community, we are surrounded by people we know and people we just met who have embraced our children and community and have committed their time, energy and resources to help us find a cure for cystinosis. Simply put, we could not have achieved what we have without the support of others.

It has taken all of us uniting for a common goal – to find a cure – to ensure that the research moves forward. We have an abundance of hope because our researchers and scientists have dedicated their lives to finding a cure for our children. Without the plethora of research we fund, we would have no reason to believe that better treatments and a cure could be found. CRF is the only foundation in the world funding millions in new research grants every year. We are the lifeline that keeps the cycle of cystinosis research community dynamic and thriving.

A NIGHT TO REMEMBER!

On April 18 more than 430 people joined us for the 2015 Natalie's Wish Celebration. It was truly a night to remember, in part because of the outstanding entertainment provided by a 10-member male a cappella group *Straight No Chaser*. An auction, which included lavish vacation packages, concert tickets and private dinners in addition to handpicked cases of fine wines, raised $384,000. Donations received prior to the event, in addition to cash raised during the evening, brought the evening's total to $2.32 million, exceeding last year's record.

By far the most special part of the night was having 45 cystinosis patients and their families from throughout the United States, Canada, Sweden and Colombia, in attendance at the event. The families have worked in their own communities to raise funds to support research. Our community is small but we are strong and determined. As in year's past, prior to the Natalie's Wish event, the families attended a three-day conference where they met with key CRF researchers and formed lifelong friendships with other families.

This year, Jeff spoke from a father's perspective about having a child with cystinosis. Jeff recalled that despite two tours of combat as a Marine Corps officer in Vietnam – he was unprepared for the "devastating" news of Natalie's illness when she was seven months old. Natalie's medical issues began as an infant when she was diagnosed with a rare tumor growing on her liver. The tumor was surgically removed along with half her liver by doctors who were unfamiliar with cystinosis – it was just the beginning of Natalie's life with cystinosis. We have reprinted Jeff's talk in the magazine (page 18).

RESEARCH AT THE FOREFRONT

Since 2003, the year Natalie made her birthday wish, ***to have my disease go away forever,*** and the year the foundation began, we have created a thriving research community. CRF is the largest fund-provider of cystinosis research in the world. CRF aggressively and strategically invests your donations, issuing grants to the best and brightest researchers in the world.

Since the beginning of the foundation, and directly because of your financial support, we now have the first donor stem cell transplantation trial open at UCLA; we funded every bench and clinical trial that led to the discovery of a delayed-release medication, which was approved by the FDA in 2013; and we have learned more about the pathogenesis of cystinosis and we are on the brink of two new 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.

CRF has issued 125 multi-year research grants in 12 countries and CRF-funded researchers have published 56 articles in prestigious journals. CRF "seed" money has been leveraged by four NIH grants totaling over $3.5 million to two CRF-funded researchers. As a result of your support, we have funded researchers whose work has exponentially increased the breadth and knowledge about cystinosis and as a result of that knowledge, new discoveries have been achieved. There are two significant research advancements since last year.

Nanotechnology for Corneal Cystinosis

CRF has been funding research that is focused on nanotechnology as a potential new treatment for corneal cystinosis. Corneal cystinosis is the painful eye condition that causes photophobia and sometimes blindness as our children reach adulthood. Although we have a treatment for corneal cystinosis it is rigorous, requiring hourly doses of medicated eye drops. Because the eye drops can be painful and the protocol demanding, compliance is difficult. Dr. Jennifer Simpson at the University of California, Irvine, and Dr. Ghanashyam Acharya at the Baylor College of Medicine, have been collaborating on research to find a better treatment.

They have developed a nanowafer that can be loaded with medication to treat corneal cystinosis. The nanowafer is placed in the eye and as it dissolves, the medication slowly releases, treating the eye for hours. If successful, the nanowafer will be a vast improvement over the current treatment. *We are excited to report that CRF has obtained the license for the cysteamine-loaded wafer. Owning the license will allow us to drive the research forward*

with the ultimate goal of a new FDA-approved treatment for corneal cystinosis. This is a multi-year project but we have an excellent team in place to ensure that we will reach our goal. We will be filing an Investigational New Drug application with the FDA within a year.

Stem Cell and Gene Therapy

Other news relates to the cure and comes from Dr. Stéphanie Cherqui's lab at the University of California, San Diego. We are very close to the first clinical trial for cystinosis using stem cell and gene therapy. Dr. Cherqui has been working closely with the FDA for several years and she is ready to move to a human clinical trial. With good news from the FDA, Dr. Cherqui formed the Stem Cell and Gene Therapy Consortium that has initiated the design of an autologous stem cell transplant treatment to be used for cystinosis. The consortium includes experts in the field of nephrology, neurology, endocrinology, gastroenterology, ophthalmology, bone marrow transplantation and gene therapy. Dr. Cherqui believes that we will have the trial ready for the first patient in 2016! If it works, the treatment will potentially cure this disease.

And beyond the world of cystinosis, CRF-funded researchers are helping other diseases. Cystinosis research has helped advance potential treatments for more prevalent and well-known disorders and diseases such as Huntington's disease and NASH – a progressive liver disease. There is great potential that CRF stem cell research will help other corneal diseases, kidney diseases and genetic diseases with systemic defects similar to cystinosis.

Your support of cystinosis research extends far beyond the cystinosis community and all of the foundation's expenses of are underwritten so that 100 percent of every dollar goes to research.

We are on the brink of new treatments, yet cystinosis remains an incurable, progressive disease – we are close to the cure, but we are not there yet. The stem cell clinical trial and the nanowafer project must be funded. Clinical trials take time and are expensive but with your continued help, we will make these new treatments a reality.

Thank You for Being Part of Our Journey

I pray every day that Natalie and the other children and adults with cystinosis will have a life free of cystinosis – no medications, no pain, no hospital visits and blood draws, no worries about life expectancy. We are extremely 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 all of you by our side, there is no doubt we will find the cure for our children.

We have accomplished extraordinary goals and we are within reach of new treatments that will change the course of cystinosis. With your support, we will continue to fund the best and the brightest researchers in the world. Thank you for supporting cystinosis research, for standing by our side and letting us lean on you when we needed it most.

With heartfelt thanks,

Nancy and Jeff

Dear Friends and Family:

Natalie (r) with her sister Alex at the Hope Lights Our Way Gala in April 2015

Last August, I started graduate school at the University of Southern California in the field of social work. I just completed my first year and will have my Masters in Social Work in May 2016. Although the work is intense, I am very happy I chose to get my master's degree in social work. I have been learning about other cultures and how being in the field of social work can greatly impact many lives. I have only been at USC for a year, but I have learned a lot. I have grown personally as well as professionally.

This year my internship involved working with families who have open DCFS (Department of Children and Families Services) cases. The Family Preservation program that I was involved with over the past nine months has really brought my own values into perspective. Very few people have what I have: an amazing support network and unconditional love from my parents, family, and friends.

In the fall, I will be interning at the Office of the Federal Public Defender in Los Angeles and learning about the criminal justice system. I am very excited about the opportunity to work at the Federal Public Defender's Office and look forward to the challenges this new position will provide.

I now know how important family is and how blessed I truly am; I cherish every moment with them. I am blessed with such an incredible and supportive family – I have learned about the true meaning of a community and I feel very fortunate to be surrounded by so many remarkable people. The cystinosis community has taught me how important it is to give back to others, and now it is my turn, as a social worker, to give back to those who are not as fortunate as I am.

At the end of the day, I have chosen to live my life to the fullest and live my life as if I did not have this lifelong and incurable illness. I will always be optimistic about my life and I will never let this disease defeat me.

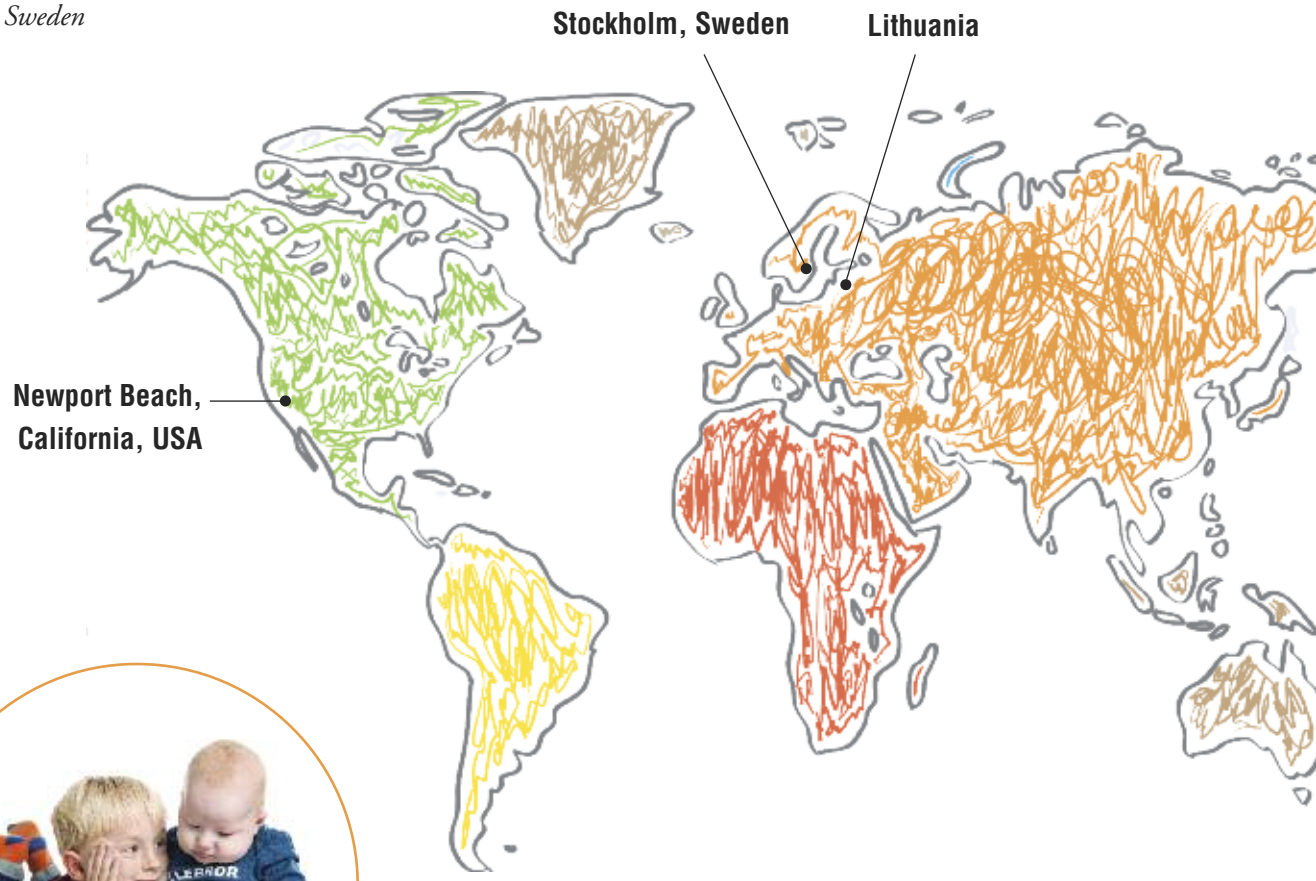
When I spoke at the Natalie's Wish event on April 18, I was surrounded by a sea of people who care about the cystinosis community. Every year I am always overwhelmed by the turnout for the Natalie's Wish event. The cystinosis community is small, but very BIG in so many ways.

Words cannot express how meaningful it is to me to know how dedicated you all are to CRF and to finding a cure. It means so much to me as well as to all the others who suffer from this disease. My parents, the doctors and, most importantly, the cystinosis community have made a life FREE of cystinosis more possible than ever. I want to thank you for your generosity, compassion, and, most of all, for your determination to make my wish come true.

Love, Natalie

Cystinosis Friends from Far and Near

By Annica Schröder, Karolis' mum, Stockholm, Sweden



Seven-year-old Karolis with his six-month-old brother, Linus

We are the Schröder family of Stockholm, Sweden. Our family of six consists of Annica (mum), Roger (dad), Andreas (21 years), Joakim (20 years), Karolis (7 years) and Linus (6 months).

Karolis was born in Lithuania, and at the age of nine months, he was diagnosed and treated for tubular acidocis. We adopted him when he was 4 ½ years old, after a year-long process. We were immediately contacted by the kidney section for children at Karolinska Hospital in Huddinge, Stockholm. Doctors at the hospital wanted to confirm Karolis' diagnosis and started performing a lot of tests.

After several months, our lives were changed completely when the doctor told us that Karolis had a rare disease called cystinosis, not tubular acidocis.

We had never heard of cystinosis, so we were very worried. The doctor told us a little about it and described the new treatments Karolis would need to start. Our lives were turned upside down. Would we now lose Karolis after finally getting him home?

We googled "cystinosis" to learn more about it and try to understand our new situation. Here we were with a new young son who we were just getting to know. Karolis didn't speak our language yet, but we had to explain to him that the doctors needed to do more tests and that he had to take more medicine than ever before. Somehow we made it.

When Karolis was diagnosed, he had a cystine level over 13. Today, it is down to 2.5, which is still too high. We have had to increase the Cystagon® very slowly because Karolis had an allergic reaction when he started taking it. We still cannot increase the dosage too quickly and risk a new allergic reaction.

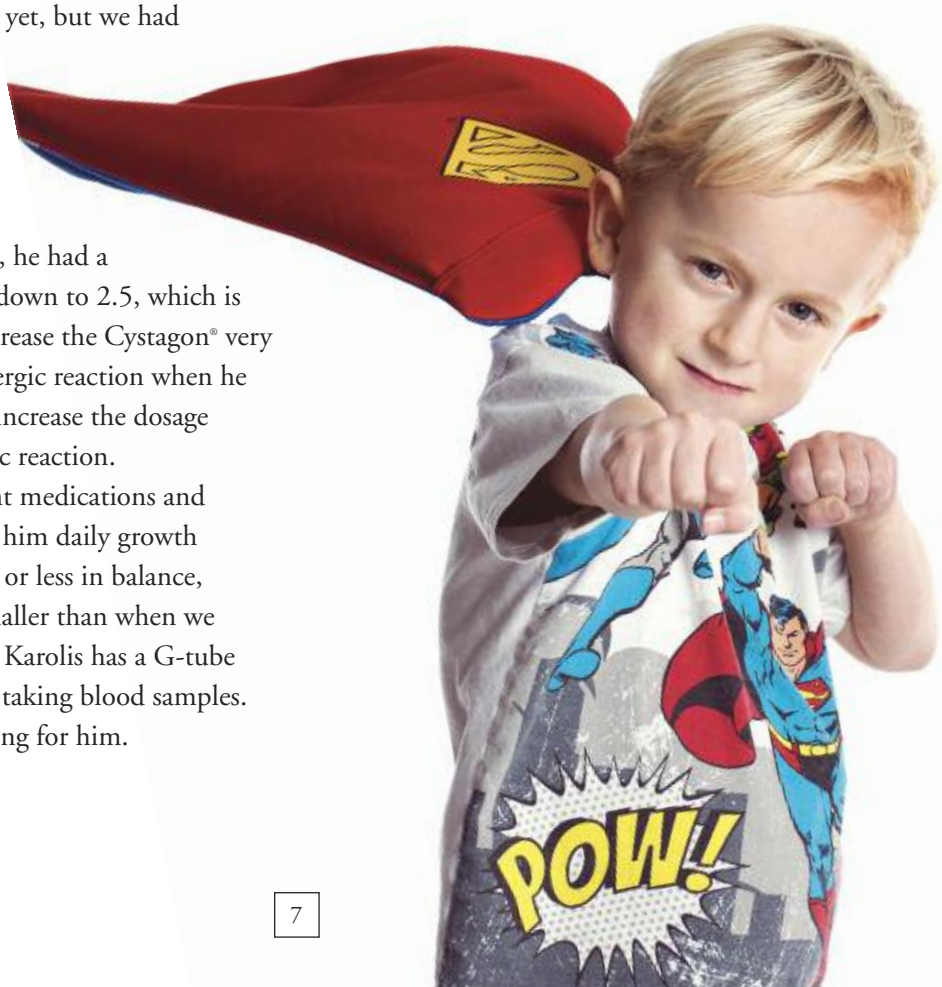
Karolis now takes 10 different medications and eye drops every day. We also give him daily growth hormones. All his levels are more or less in balance, and the crystals in his eyes are smaller than when we met the eye doctor the first time. Karolis has a G-tube in his stomach and a catheter for taking blood samples. Both of them help us a lot in caring for him.

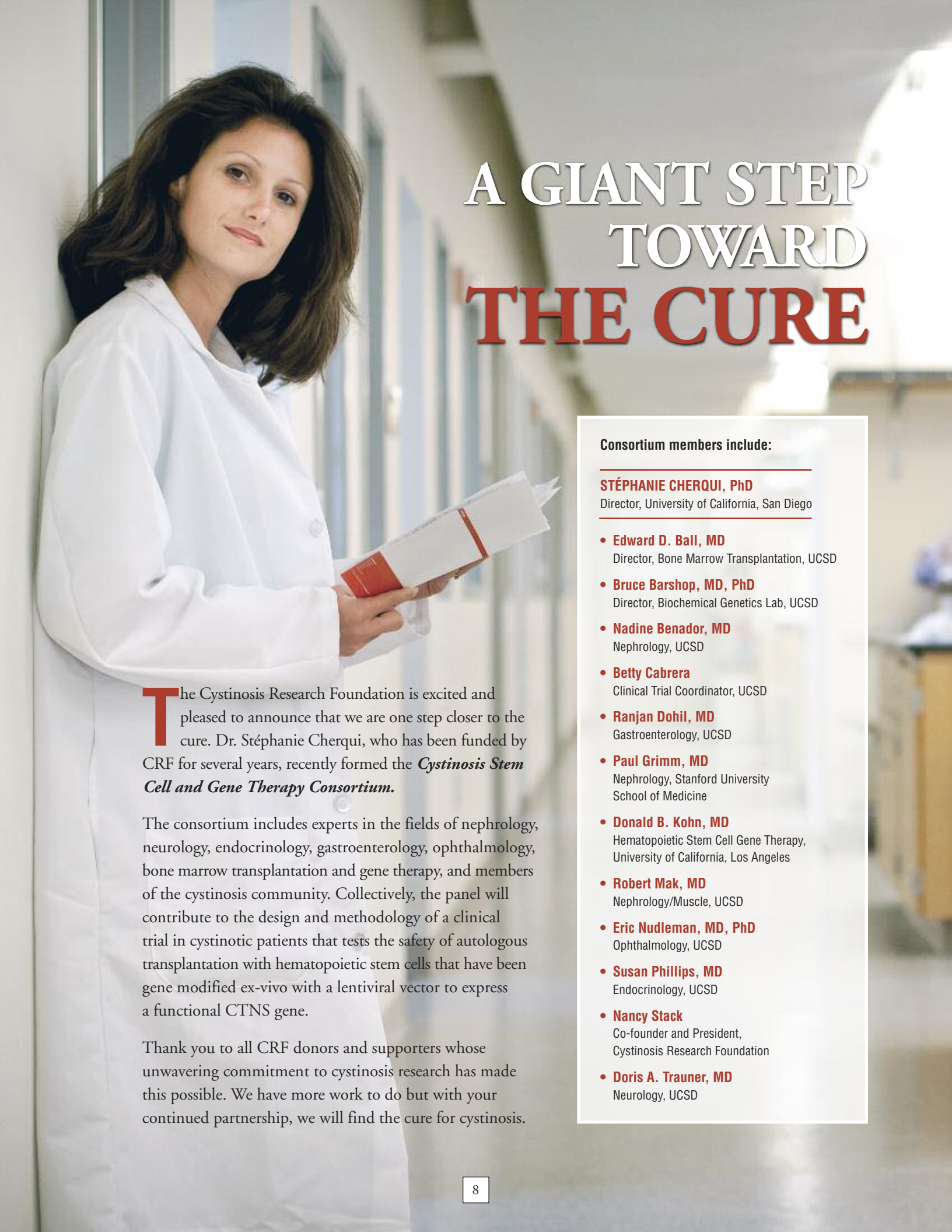
Karolis now understands that he has a rare disease and that he needs to take his meds every day for the rest of his life. He hates his illness and tells us every day that he hopes that he can get well soon so he can stop taking his meds, and get rid of the catheter and G-tube – and be like his friends. He feels that he is different, which is hard for a 7-year-old boy.

There aren't many cystinosis patients in Sweden, so Karolis has never met anyone else with the disease. Fortunately, we are members of a kidney organization in Sweden where he has met several kids with other types of kidney conditions, which helps him accept his situation.

In all other respects, Karolis is just like any other young boy. He attends school, plays with friends, practices soccer and he loves to make his little brother laugh.

We hope we will be able to attend the CRF Day of Hope Family Conference in 2016 and that Karolis will meet other kids with cystinosis and share his thoughts with them.





A GIANT STEP TOWARD THE CURE

The Cystinosis Research Foundation is excited and pleased to announce that we are one step closer to the cure. Dr. Stéphanie Cherqui, who has been funded by CRF for several years, 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 panel will contribute to the design and methodology of a clinical trial in cystinotic patients that tests 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.

Thank you to all CRF donors and supporters whose unwavering commitment to cystinosis research has made this possible. We have more work to do but with your continued partnership, we will find the cure for cystinosis.

Consortium members include:

STÉPHANIE CHERQUI, PhD
Director, University of California, San Diego

- **Edward D. Ball, MD**
  Director, Bone Marrow Transplantation, UCSD
- **Bruce Barshop, MD, PhD**
  Director, Biochemical Genetics Lab, UCSD
- **Nadine Benador, MD**
  Nephrology, UCSD
- **Betty Cabrera**
  Clinical Trial Coordinator, UCSD
- **Ranjan Dohil, MD**
  Gastroenterology, UCSD
- **Paul Grimm, MD**
  Nephrology, Stanford University School of Medicine
- **Donald B. Kohn, MD**
  Hematopoietic Stem Cell Gene Therapy, University of California, Los Angeles
- **Robert Mak, MD**
  Nephrology/Muscle, UCSD
- **Eric Nudleman, MD, PhD**
  Ophthalmology, UCSD
- **Susan Phillips, MD**
  Endocrinology, UCSD
- **Nancy Stack**
  Co-founder and President, Cystinosis Research Foundation
- **Doris A. Trauner, MD**
  Neurology, UCSD

Tina Flerchinger:

MY INSPIRATION AND MY HERO

By Julie Flerchinger, Tina's 24-year-old cousin, Bellingham, Washington

Family has always been a cornerstone in my life. When I first learned that another cousin would be added to the Flerchinger family, I was excited, but I had no idea how much of an impact she would have on me. In November 2003, my family and I headed to Clarkston, Washington, to meet Tina.

As soon as I held her, we had a special connection. I knew that God brought Tina into my life for a special purpose. I had no idea that she was to become my inspiration and my hero.

Tina was diagnosed with cystinosis at 17 months. I had never heard of this disease and my mind raced with questions. What is cystinosis? How could someone so small and precious be affected by it?

I watched Tina refuse to eat, throw up when she did, drink gallons of water, lose weight, and continue to struggle with her health. I was told that there was no cure for cystinosis and that it could take her at any moment. I refused to believe those words.

When we went to visit, I spent every moment with her. Everyday was precious, and I did not want to waste a single second. As I watched her grow, I would sometimes forget that cystine was accumulating in her body and affecting her organs. Her face lit up when we played, and laughter would ring through the air. She embraced life with such enthusiasm that I would almost forget she was sick.

When I finished reading her a bed time story one night, she asked if I would say her prayers with her. After finishing praying for people in the family, she asked me to pray for her. I told her, "Of course! I pray for you every night." She went on, "I know I am sick, and I want to feel better so my family doesn't have to worry."

That is when it hit me. Tina knew she was very sick, yet she was not concerned with her needs as much as she was worried about her mom, dad and sisters.

Tina has always been and always will be my hero. I have never met anyone so strong, loving, caring, happy and full of life. On the outside, no one would know that she takes many medications every day to stay alive. She has taken more than 47,000 syringes of liquid medications and 58,000 pills. I have been with her many times when she was taking her meds, and not once have I heard her complain about them. Tina simply smiles, takes her medications, and then grabs my hand so we can go back to playing.

She is strong, brave and determined. Cystinosis will never keep Tina from sharing her love and happiness with those around her. She inspires everyone she meets. Tina reminds me every day how precious life is; what the important things are in life; and how to make a smile and laughter contagious to those around you.

Tina's inspiration and impact on our lives led to the creation of *Tina's Hope For a Cure,* a nonprofit foundation that raises money for the Cystinosis Research Foundation. Together, we are working to find a cure for cystinosis.

Tina has touched our hearts and we work to embody her strength, bravery and determination in order to raise awareness of this rare disease and to find a cure. We will not give up until Tina and others who are affected by cystinosis know what it is like to be healthy.

I would like to address the importance of remaining compliant when it comes to taking all your medications – and the important lesson I learned the hard way.

A HARD LESSON HARD LEARNED

By Tanner Edwards, a cystinosis patient from Fort Collins, Colorado

Tanner Edwards with his mother Traci Gendron at the 2015 Natalie's Wish Gala. Traci donated her kidney to Tanner on October 16, 2014

When I was younger my mom always made sure I took my medicine. Since I'd taken medication my entire life, taking it was normal to me. When I started high school, it became a huge inconvenience. I hated having to stop what I was doing to take my meds, especially if I was playing sports with my friends. I also got tired of having a stomachache when I went to school, and I was jealous because my friends never had to worry about taking medicine.

So one day I skipped a dose of medication and I didn't feel any different. That turned into more skipped doses, until I just stopped taking my medicine all together. I was 14 years old and I figured I knew everything. I thought to myself, "I don't feel sick. Now I'm just like everyone else."

That summer, I received my first transplanted kidney from my dad. It was a planned transplant because my creatine was rising. I was never on dialysis – putting someone so young on dialysis if it was unnecessary didn't make sense. The transplant went well and all I remember was everyone asking me if I felt better. I didn't feel sick to begin with, so my answer was always, "I don't feel any different."

But now, I had to take new medicines, such as antirejection meds and pills, so I didn't get infections. About two years after my transplant, I stopped taking my transplant medicine consistently. I would take it, then skip a couple of doses, and then start it again. I was young and stubborn, and very lucky that my kidney didn't fail immediately. My mom would be in the kitchen with me before school and I would pretend that I was taking my medicine. Instead, I would hide it in my mouth or pocket, and then hide it in my room or throw it away when I got to school.

This continued when I started college. Now that I was on my own, the last thing I thought about was taking medicine. I was more concerned about going out with my friends and having fun.

College wasn't really for me, so I got a job. Things were going well until I started having bad headaches and became very moody for no reason. I figured it would all go away and never really thought much about it until one day I had such a severe headache I felt like crying and told my mom I needed to go to the hospital. My blood pressure was 260/168 and I was lucky I was still alive. My creatine was 9.2 and I was told that I would need a new kidney and was put on dialysis.

I had my first tranplanted kidney for 12 years, which is a miracle since I took horrible care of it. And I had wasted my dad's kidney, which I feel bad about. I was in the hospital for 24 days and I felt terrible. I was then on dialysis for two years, which nobody should have to experience because it's not living, it's just keeping you alive.

Younger kids who take these medications should be taught what these medicines do for them, so they understand how important it is.

I received my mom's kidney on October 16, 2014 and have felt amazing ever since. **I will never miss a dose of medicine again.** Going on dialysis actually woke me up and showed me how important it is to take your meds. I never want to feel that bad again.

I believe that younger kids who take cystinosis medications should be taught what these medicines do for them, and why they are so important to them. I always felt that when a doctor told me to take all these pills and didn't take any himself that he didn't know how I felt. Education is very important with younger kids. I think that if I had heard all of this from an older cystinosis patient, I might have listened and I might still have my first kidney.

I would gladly talk to anyone who is struggling with being compliant because I can relate to him or her, and think I can help them out.

Today, I feel great and have a great outlook on life now – and I will never miss any more doses of my meds.

Tanner with his mom, Traci, who donated her kidney to him

Becoming a Kidney Donor

Many people become donors because of a family member or friend in need. However, non-directed or anonymous living donations can help those unable to find a match within their support community.

The National Kidney Foundation – www.kidney.org – can provide detailed information and resources, but here's an introduction to the process of becoming a living donor:

- Contact your local hospital or dialysis center to start the process. The National Kidney Foundation has a search engine to help locate a center near you. The first step to getting on the list of potential donors is an appointment to complete paperwork.
- You will be asked to provide proof of your age and other information. Sometimes you'll be assigned to a counselor for an interview to discuss the process and commitment.
- A doctor will take your complete medical history. Blood will be drawn to type and match to six tissue types necessary for an organ to be the best match. Additional tests will be performed to check if your kidneys are functioning properly.
- If you are approved, you will be put on a donor list. When and if your kidney is needed, you will receive a call, giving you the chance to provide someone with the gift of life.

Growing up I always remembered people saying how fast time flies; that moment struck me for the first time when we started packing for our fourth Day of Hope conference.

Time Flies when you're having fun

By Chad Little, Olivia's dad, Saugeen Shores, Ontario, Canada

In a blink of an eye Olivia is five years old, Harper is almost two, and we have been a family living with cystinosis since Olivia was 18 months old. I still remember bringing Olivia home from the hospital for the first time on a snowy day in December with my wife Erin. Olivia was so little and perfect in every way.

Erin and I talked about all the things Olivia's future would hold. Would she attend university or not? If she did go to school what kind of degree would she get? What kind of career path would she take? But never once did it cross our minds to discuss at what age would her disease be cured.

Our focus has shifted from dreaming of what the future would bring for Olivia to what kind of future we are going to make for her. A future of not having to travel to Sick Kids Hospital for regular appointments; a future of not prepping medication aka "vitamins" for the week; a future of not living by a clock to ensure that her six hour doses are met; a future of not chasing her every hour to administer eye drops; a future of not waking up to change diapers, bed pads, sheets and pillow cases; a future where being spontaneous doesn't include days of planning and packing to ensure that we "have everything."

On a long road trip to northern Minnesota from southern Ontario, the Liv-A-Little Foundation was born. We couldn't stand by and watch the efforts of CRF and the other families work so hard, while we did nothing.

The support we have received from our family and friends, the community, and even complete strangers, has been overwhelming to say the least. We are still astonished that in a town of 12,000 people we raised more than $130,000 in two years and we are on track to raise $100,000 this year.

The people we have met and the places we have gone, would never be part our lives without cystinosis.

I still struggle seeing my little princess having to be on a regimented medication schedule just to keep her alive.

I cannot shake the memories of those nights waking Olivia up from a deep sleep just to pin her down so that Erin could administer her midnight dose.

I am worried about future health implications, such as kidney transplants or side efforts from her current medication.

I battle with the thoughts of her medicine compliance in the future.

I dread the day she realizes she has a disease or figures out that Harper (her little sister) does not need any medication.

I hope I give Harper the love and attention she deserves.

I wish I could just trade places with Olivia and give her my health and that I would have cystinosis for her.

More than anything, I want Olivia to be Olivia and not Olivia with cystinosis because right now she is only five years old and has no idea.

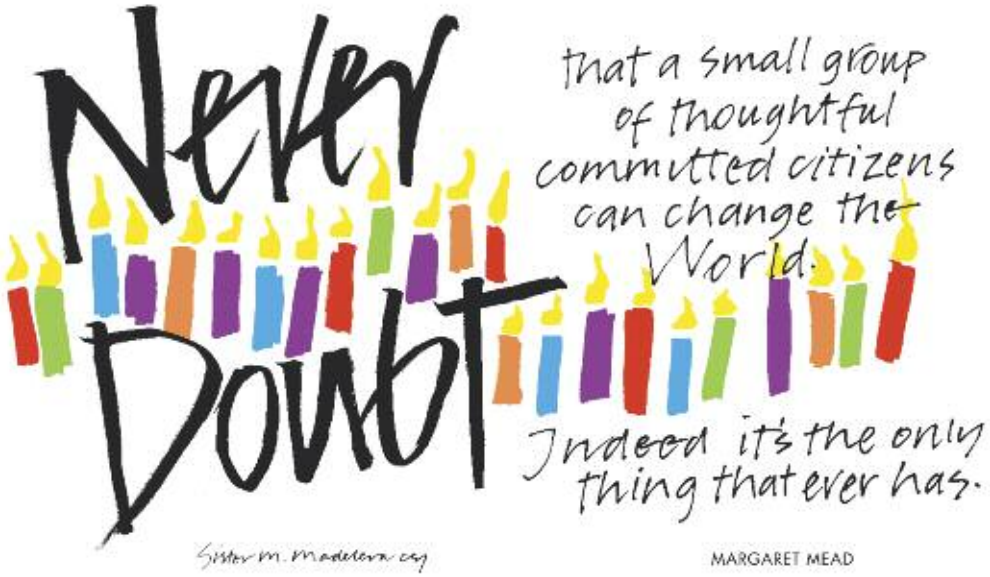
She has already made such a powerful impact and contribution to our lives and enriched our family connection.

I can't express how grateful I am to have such a great partner in life. My wife, Erin, takes the brunt of the work and has adjusted her life to become a full-time caregiver to Olivia.

Thanks to Nancy and Jeff, CRF, our families, friends, community and all the other families who give us hope.

For our daughter Olivia, our greatest inspiration.

Olivia, Erin, Chad and Harper at the 2015 Day of Hope Family Conference



In 2014, donors from around the world helped CRF raise $3.5 million for cystinosis research.

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

Jenna & Patrick Partington
$226,900

Holt Grier
$178,171

Tina Flerchinger
$124,371

Henry Sturgis
$120,100

Jake Krahe
$102,390

Hadley Alexander
$67,179

Landon Hartz
$31,545

Bailey DeDio
$28,418

Olivia Little
$26,833

Gabbie Strauss
$26,000

Joshua Clarke
$25,000

Mary Head
$17,196

Andrew Cunningham
$16,466

Nicole Hall
$15,544

Abbi Monaghan
$15,495

Caleb Gowan
$6,105

Sam and Lars Jenkins
$5,939

Camden Sanders
$4,500

Tanner Edwards
$3,986

In memory of Sarah Melang: $14,175

Keegan Manz: $9,561

In memory of Grandmother Nora 'Dolly' Kugler, Morgan Peachman: $4,420

Amanda's Hope for a Cure, Amanda Kuepfer: $2,273

In memory of Nathaniel Wagler: $885

Chandler's Chance, Chandler Moore: $534

Mika Covington: $527

How far that little candle throws his beams! So shines a good deed in a weary world. SHAKESPEARE

***Today, CRF-funded research has the potential to help millions of people worldwide who have more prevalent and well-known disorders such as Huntington's disease and NASH, a fatty liver disease, as well as other rare lysosomal storage diseases, kidney diseases and corneal diseases.**

Totals for year ending 12/31/14

WOW! What a Night!

On Saturday, April 18, more than 430 guests packed the ballroom at the Balboa Bay Resort for the 2015 Natalie's Wish Gala. The huge crowd helped the Cystinosis Research Foundation break last year's record by raising more than $2.35 million for cystinosis research.

Guests traveled from all corners of the world to join this annual celebration of milestones CRF has already achieved – and to cheer the possibilities that lie just ahead on our journey to find the cure. Forty-five patients and their families from as far away as Sweden and Colombia to celebrate a vastly improved quality-of-life because of CRF- funded research.

Guests were awed from the minute the ballroom doors opened, casting an enchanting glow that perfectly matched the theme of this year's event: *On the Road to the Cure, Hope Lights Our Way.*

That was only the first of many dazzling moments during this inspiring night. Each year, Nancy Stack finds an enchanting group to perform at the event, and this year was no exception. Straight No Chaser, a 10-member male a cappella group, wooed the audience with their charisma, their harmony, and their obvious and heartfelt appreciation for Natalie and her now-famous wish.

As always, the night's highlight was a live auction that raised $384,000 and included lavish vacations, concert tickets, dinners and exceptional wines.

The evening's most anticipated moment came when Jeff Stack, Natalie's father, addressed the audience. Jeff recalled that even his two tours of duty in Vietnam as a Marine Corps officer did not prepare him for the "devastating" news of Natalie's illness when she was seven months old. His heart-wrenching story mesmerized the audience, while rallying them in the fight against this terrible disease.

While there is still much work to be done in the fight against cystinosis, the generosity of Natalie's Wish attendees and CRF supporters from around the world brings us closer to a cure every day and gives hope to the children and adults with cystinosis.

The Stack family would like to thank everyone who helped make this year's event an unparalleled success.

CRF President Nancy Stack welcomes guests to the 2015 Natalie's Wish Celebration

CRF Board Member Michael K. Hayde, Sue Werner and CRF Board Member John Hagestad

Longtime family friends and CRF supporters Bob and Christyne Olson

10 singers
20 cystinosis researchers
430 guests from around the world
450 donors
2,000 children and adults with cystinosis
$2.35 million raised for research

1 heartbreaking wish
"to have my disease go away forever"

Special Stack family friends Cindie, McCallister and Bert Selva

CRF Board Member Bruce Crair and his wife Cathy, with Board Members Traci and Tom Gendron

High-spirited bidding during the live auction, which raised $384,000 for cystinosis research

Jeff Stack has worn many hats and spoken to many impressive audiences through the years, but none more important than when he told Natalie's story of courage and hope at the 2015 Natalie's Wish Celebration. Here are the heartwarming and heart-wrenching sentiments he shared that night.

The four most meaningful events in my life are my marriage to Nancy in 1987 – tomorrow we will be married for 28 years – and the births of my three daughters: Katie in 1974, Alex in 1989 and Natalie in 1991.

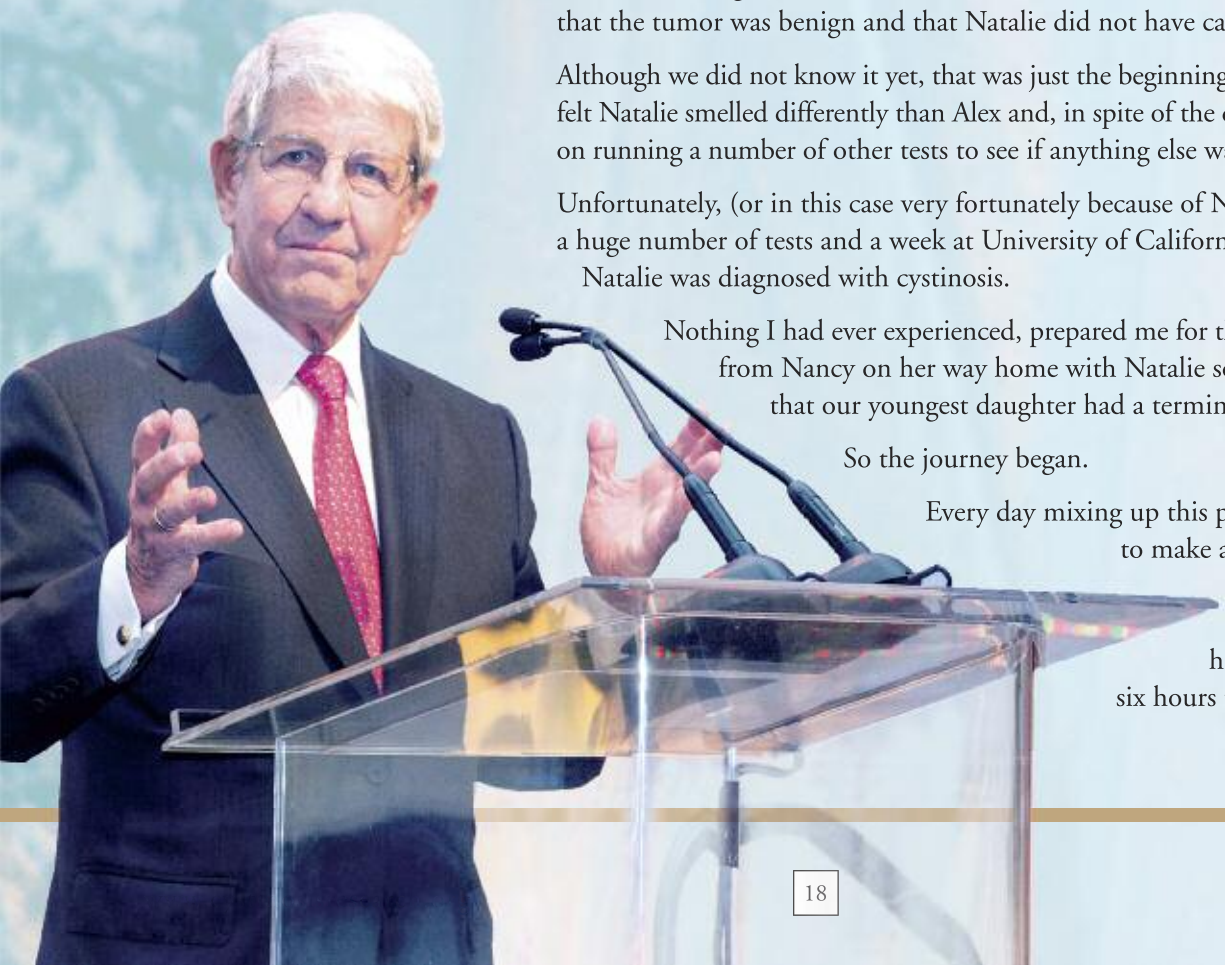
As many of you know I am a Marine. I spent two combat tours in Vietnam and unfortunately saw a great deal of death, destruction and mayhem.

Nothing in my Marine experiences, however, prepared me for that fateful call from Nancy in April 1991, telling me that she was on her way to Children's Hospital in Orange because two-month-old Natalie appeared to have a tumor and I needed to meet her there as soon as possible. We spent an agonizing afternoon going through numerous tests, MRI's, etc. only to find out that Natalie did indeed have a tumor attached to her liver.

The doctors were sure it was malignant and Natalie needed to be operated on quickly. That was on Friday and we had the longest most agonizing weekend of our lives.

THE AGONY *& The Ecstasy*

By Jeff Stack, Natalie's dad and co-founder of the Cystinosis Research Foundation



Early the next week, we spent a day in the pediatric oncology ward getting briefed on all we would have to do after Natalie's operation to combat the cancer they were sure she had. That was a devastating day for me, and I still remember it vividly!

Long story short, a day or two later, Natalie underwent a seven-hour surgery and the tumor, along with half of her liver, was removed. Miraculously it turned out that the tumor was benign and that Natalie did not have cancer.

Although we did not know it yet, that was just the beginning of our journey. Nancy felt Natalie smelled differently than Alex and, in spite of the doctor's advice, insisted on running a number of other tests to see if anything else was wrong with Natalie.

Unfortunately, (or in this case very fortunately because of Nancy's vigilance) after a huge number of tests and a week at University of California, San Diego hospital, Natalie was diagnosed with cystinosis.

Nothing I had ever experienced, prepared me for the next fateful call from Nancy on her way home with Natalie sobbing uncontrollably that our youngest daughter had a terminal illness.

So the journey began.

Every day mixing up this powdered medicine to make a vile tasting and smelling liquid, which Natalie had to take every six hours to stay alive.

Every night for years, waking Natalie up at 11 p.m. and 5 a.m. and holding her down while she screamed and cried so we could give her the medicine to help keep her alive.

Making numerous trips to the emergency room because Natalie wasn't doing well or had fallen due to balance issues and we were concerned she had a concussion.

I could go on and on, but I think you get the picture – we were struggling every day with Natalie's disease and all the attendant problems, sorrows and troubles this dreaded disease caused us.

I will say this – Natalie had an indomitable spirit through it all no matter what happened! We called her our "littlest Marine" and she lived up to that title every day!

The good news is that there is a bright side to this story and many of you know all or part of it.

Twelve years ago in 2003, we (but it was really Nancy) decided we had to do more to combat cystinosis than simply struggle in our own family.

We decided to go public to start the Cystinosis Research Foundation to raise money for research to find better treatments and a cure for cystinosis.

We did this with great trepidation because we are basically private people and did not want people feeling sorry for us, nor did we want to burden them with our problems.

Some of you here tonight were there for our first fundraiser and showed us then what wonderful, caring and supportive people you were!

What has happened over the past 12 years is nothing less than miraculous!

We have raised over $29.3 million for medical research. Thousands of people around the world have donated money and embraced this cause.

As Nancy mentioned, we have had incredible success in our research and made major breakthroughs in finding better treatments and ultimately a cure for cystinosis.

Amazingly along the way there now appear to be applications to several other diseases as a result of our research.

Most importantly, we have received an overwhelming outpouring of love, generosity and deep caring from not only all of you, but from many, many others around the world.

We have had an opportunity to watch Natalie grow and succeed – graduating from grade school, high school, and last year from Georgetown University. Now she is in graduate school at USC and doing very well.

Natalie's indomitable spirit has given me strength and courage over the years and made me a far better person.

Natalie, you are still my "littlest Marine!"

We have discovered this is an incredible world filled with extraordinary people who have become our family. This journey has often been hard and incredibly daunting, but because of your generosity, your spirit and your overwhelming love, it has become joyful and most rewarding.

This dreaded disease has brought all of you into our lives and your love and support have deeply enriched our lives.

Thank you for listening, and thank you for your overwhelming support!

Hope Lights Our Way

2015 NATALIE'S WISH CELEBRATION

We are grateful to each of you for lifting the Cystinosis Research Foundation to heights greater than we could have ever imagined.

With your help we are moving ever-closer to making Natalie's wish – *to have my disease go away forever* – a reality.

We still have much to do but your generosity gives real hope to the children and adults in the cystinosis community who desperately need our help.

$500,000 and above

Traci and Tom Gendron

Nancy and Geoffrey Stack Family Foundation

$100,000 – $150,000

Laura Khouri and Michael K. Hayde

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

$50,000 – $70,000

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

Susan and Thomas F. Moran

Chris and Bill Thormahlen

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

$20,000 – $30,000

Nicole Frank and Bob Barth

Deborah and Larry Bridges

Caleb's Cause, *in honor of Caleb Gowan*

Chance for Chase, *in honor of Chase Chodakowsky*

The Allan Hunter Family

Suzu and David Neithercut

Mary and Peter Rooney

Rosemary and Bruce Rose

Today there is no cure,

$15,000–$19,999

Anonymous
Nelda Arnold and W. Dean Henry
Mr. and Mrs. Chris P. Dialynas
Ralph H. Eidem, Jr.
John Hagestad
Joshua's Journey of Hope,
in honor of Joshua Clarke
Elizabeth and Steve Rabosky
The Redfern Gallery,
Beverly and Ray Redfern
Sam and Lars' Hope for a Cure,
in honor of Sam and Lars Jenkins


$10,000–$14,999

Kim Azar-Shepperd
Kevin Covey
Gary Hunt
Mary Ann King and Alex Rose
Lincoln Property Company
Jill and John Manly
Paula and Michael Murphy
Lynne and Augie Nieto
Christyne and Bob Olson
Cindie and Bert Selva
Sandy Sewell,
in memory of Sarah Melang
Dr. Jennifer Simpson and Dr. Ron Kurtz

$5,000–$9,999

Allen Matkins
Wendy and Rick Aversano
Joan and Donald Beall
Shelley and Phillip Belling
Berle Family Trust DTD,
Sharon and Rod Berle
Cathy and Bruce Crair
C.C. Pascal Charitable Foundation
Kristin and Aram Chahbazian
DLD Insurance Brokers, Inc.,
Dana Dowers
Gerson Bakar & Associates
Colleen and Jack Hammond
Heidi L. and James D. Harper, Jr.
Irvine Company Community
Development
Peter Kompaniez
Janice and John Markley
Sue Ellen and Paul O'Connor
Raptor Pharmaceuticals
The San Francisco Foundation,
Rob Wagner and Mark Kroll
Silverline Construction
Zoe and Don Solsby
Sullivan Curtis Monroe Insurance
Services, LLC
Tejon Ranch Company
Jill and Larry Tucker
UC Irvine Health
US Bank
VCA Structural
Linda and Joseph Zeller

$2,500–$4,999

Abbi's Sunglass Day,
in honor of Abbi Monaghan
Michael Bissell
Dr. Michele and Ed Carpenter
Judy and Dave Christensen
Connie and Ken Coatsworth
Barbara and Edward Geraghty
Lori and Richard Glucksman


Grandpoint Bank
Barbara Grimm Marshall
Beth Anne and Don Haarer
Hearts for Hadley,
in honor of Hadley Alexander
Hillgren Family Foundation Fund,
Lisa and Bradley Hillgren
The James M. Peters, Jr.
Revocable Trust
Dr. Stephanie Lu
Ellen and Scott McGuirk
Natalie and Thomas P. Moran


Dr. Sue Murahata and Dr. Dale Kistler
Ceil and Richard Nelson
Nine Dots Fund, RL and
David Peters Family
Teresa and Kevin Partington, *in honor of Jenna and Patrick Partington*
Lisa and Chris Payne
Schaefer Real Estate Partners, LP
Tina's Hope for a Cure,
in honor of Tina Flerchinger
Sara Elise-Volbert and
Terrance Volbert

$2,000–$2,499

Lan and Dr. Charles Bolus
Carolyn and C. Preston Butcher
Tami and Bob Cluck
Kim and Dick Crawford and Family
David E. Martin Architects
Brenda and Brian Deeley
Family Foundation Fund
John Ginger
Park Newport Apartments
Shannon and Byron Tarnutzer
TCA Architects, Inc.

$1,000–$1,999

Eric Affeldt
Hope Aldrich and Michael Jeffers
Marjorie and Ed Alvarez
Amanda's Hope for a Cure,
in honor of Amanda Kuepfer
Yvonne Anderson and Steven Betts
Bank of the West
Barcelona Apartments
Dr. and Mrs. Bruce Barshop
Sandy and Dr. Stephen Bell
Lesley and Dr. Alan Beyer
Jean Blum and Carl Lang,
in honor of Jackson Blum-Lang
Buck Owens Production Company, Inc.
Mary V. Buckingham
Call & Jensen
Chapman, Glucksman, Dean,
Roeb & Barger
Maidy and Col Robert Coates,
USMC (Ret)
The ConAm Group
Cystinosis Awareness & Research
Effort, *in honor of Gabbie Strauss*
Heather and John Densmore
Draper Family Foundation Fund,
Priscilla and Ranney Draper
Linda and Kris Elftmann
Wendy and Chris Ferraro
Max L. Gardner
Maggie and Ehsan Gharatappeh
Stephanie and Jim Gilly
Dr. Karen Grimley
Leslie and Larry Jenkins,
in honor of Sam and Lars Jenkins
Chris and Roger Jolicoeur
Gayle and Roy Jones
Chris and Brent Justice,
in honor of Hunter and Shyann Pugh
Linda and John Kensey
Kilimanjaro Climb,
in honor of Katie Ahnen


Valerie and Bill Kircher
Anne and John Kittleson
Dr. Jana and Nathan Kraulik,
in honor of Caleb Gowan
Lots of Love for Landon,
in honor of Landon Hartz
Karen and Tom Linden
Frank Martell
Sherri Martin
Scott McCarty
Carolyn and Gary McKitterick
Miramar Venture Partners
Angela and Bill Montgomery
Barbara and Pat Moore
Music for Mary, *in honor of Mary Head*
Pacific Premier Bank

but because of you, they have hope!


Kathy and Steve Prough
Race for Nicole, *in honor of Nicole Hall*
Denise and Patrick Russell
Keiko Sakamoto and Bill Witte
Elaine and Dr. Jerry Schneider
South Coast Plaza
St. Moritz Apartments
Sherry and Rick Stinehart
Cindy and Al Stokke
Tanner's Time for a Cure,
in honor of Tanner Edwards
The Taylor Family Trust,
Colleen and Michael Taylor
Cassie and Jim Thomas
Venture Tax Group LLP
Wear Your Rare Campaign,
in honor of Ethan Bartkowski, Keegan Manz and Chandler Moore
Ann and Marc Winthrop

$500–$999

Amber and Scott Alexander,
in honor of Hadley Alexander
Lynn and Art Barrett
Laurie and Randy Boehme,
in honor of Zylar Smethurst
Sherry and Tim Boucher,
in honor of Bailey DeDio
Wayne Brander
Elena Cappadona and
Stephen Stambaugh
Diane and Jim Connelly
Cindy and Douglas Crocker
Sandra and Wm. Gary Crouch
Ted Daley
Cyndee and Jon Ely
Feldman Family Trust,
Patti and Dr. Edward Feldman
Nedra Ferraro
Donna and Russell Goodman
Ruthy and David Green
Janet and John Griffith
Mary Ellen Guignon and Tyler Richards
Carl 'Chip' O. Harvey III
Donna and Michael Heiken
Hope for Holt, *in honor of Holt Grier*
Michael L. Horst
Olivia and Andy Johnson
Nikie and Matthew Le
Mack Maxwell
Andrea and Tim McGinley
Dina and James Mead
Erin and Todd Meyer
Julie and Jack Mitchell
Nancy and Mark Nimmo
Paula and George Norton
Jennie and Darrin Nutter
Bonnie and Hillar Paju,
in honor of Shannon Paju
Michele and Hector Perez
Nancy and Luu Phan
Michael Pier
Sheila and Ygal Sonenshine
Lynda and Ken Stack
Andrea and Ivo Tjan
Paula Tomei and David Emmes
Dr. Doris Trauner and Dick Stanford
Stephanie and Jerry Walker
Lois and George Wolfe

$250–$499

Joann and Henry Acuff
Nelda Arnold and W. Dean Herny
Debbie and Speedy Beers,
in honor of Zacqary Beers
Hinda and Hal Beral
Susie and Bo Burch
Mary Ellen Carter
Dr. Stéphanie Cherqui
and Fabien Faucheux
Allen Court
Joan and Robert Curtin
Susan and Peter Denniston
Lara and Peter DiCorpo
Joan and George Gendron
Carol and Ron Gillespie
Timothy Gleason
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Debbie and Greg Heath
Mildred Heaton
JPMorgan Chase Foundation,
Brian Okrent and Michael Yoo
Virginia Kimonis, MD
Ellen Kubo and Allen Kirschenbaum
Stephen Lanni
Sally and Bruce Lawrence
Kathy and Richard Leventhal
Friends of Keegan Manz
Celia McMaster
Marylyn Milburn
Liz and Bob Miller
Mulligans for Morgan,
in honor of Morgan Peachman
Ryan Patterson, *in honor of Emma and Gracie Patterson*
Dorothy Phan
Karen Shoeman and Kenneth Post
Margaret and Gen. Francis Quinlan
Zachary Rabosky
Debbie and Frank Rugani
Mari and Col Ray Snyder (Ret)
Tricia and Brian Sturgis,
in honor of Henry Sturgis
Rhonda and Craig Tudor
Julie and Justin Wilson
Shara and David Witkin
Mary Beth and Don Woods
Joan and Jim Ziegler


$100–$249

Chad Anderson
Dr. Angela Ballantyne
Bonnie and Stan Bastian
Mary Rose and John Booth
Sergio Catz, PhD
Karin and Kendall Ching
Don and Karen Cunningham,
in honor of Andrew Cunningham
Kelsey Cunningham,
in honor of Andrew Cunningham
Kristine David
Jessica and Jay DeDio,
in honor of Bailey DeDio
Gail Friedt
Kaitlin Geuss and Ed Piddington
Lisa Glass
Deborah and John Hagan,
in honor of Jake Krahe
Claire Hartzell and Nicole Groux
Cinnamon and Jeffrey Hawks
Hopes & Wishes,
in honor of Jake Krahe
David Howard
Pamela and Jeffrey Howard
Cathy and Scott Janke
Jackie and Kevin Keizer,
in honor of Shannon Keizer
Terry and Phillip LaMotte
Leslee and Lewis Levey
Sharon and Duayne Manz,
in honor of Keegan Manz
Sandy and Lt. Col. Jerry Martin (Ret)
Cathy Michaels
Laurie Mola, *in honor of Bailey DeDio*
Kris and Jack Morris, *in honor of Jenna and Patrick Partington*
Philippa Muir
Chau and Thomas Nguyen
Kaylee O'Connor
Kelly and Eric Olsen
Cassidy Penner
Betsy Reed, *in honor of Jenna and Patrick Partington*
Kari and Dr. David Richardson
Raymond Rourke
Lauren and Robert Silvernail
Caren Ureda
Marcela Vargas and Mauricio Quijano,
in honor of Marcela Quijano Vargas

$99 and Under

The Boeing Company, Vanessa Milburn
Exelon Corporation Employee
Giving Campaign, Anonymous
Cassie Keturakis,
in honor of Natalie Stack
Rosalin Sakdisri
Marjorie and Harold Zimmerman

Gifts-in-Kind

The Golden Truffle Catering/Restaurant,
Alan Greeley
J.K. Bookkeeping, Karen Dimmick
Jaeger-LeCoultre
Laura Khouri and Michael K. Hayde
Kohanakiki Club, Inc., Johnna and
Don Grant, Paula and Doug Neff
Dr. Susan and Mark Kroll
Montage Laguna Beach
Multihousing Professional Magazine,
Linda Hoffman
Noelle Marketing Group
Patina Catering,
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Nancy and Geoffrey Stack
Straight No Chaser
TPL Properties, Richard Eddy
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WITH A SPECTACULAR NEW COURSE, ORANGE COUNTY'S BEST TOURNAMENT IS BETTER THAN EVER.

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

THE RESORT AT
PELICAN HILL
NEWPORT BEACH

Mark your calendar for the 2015 Fore a Cure Golf Tournament, which will be held at the world-class Pelican Hill Golf Club on Monday, October 19, 2015.

For sponsorship information contact Zoe Solsby at (949) 223-7610 or zsolsby@cystinosisresearch.org.

Course photos courtesy of Pelican Hill

Day of Hope

CYSTINOSIS RESEARCH FOUNDATION

FAMILY CONFERENCE

Story by Stephen Jenkins, MD, Samuel and Lars' dad, Salt Lake City, Utah

Learn Laugh Share Celebrate

"For our family, the Day of Hope Conference is even bigger than Christmas!

Stephen Jenkins, MD

We just returned from Newport Beach, California, where we attended the 2015 Cystinosis Research Foundation Day of Hope Family Conference. We look forward to it all year.

The conference kicked off on Thursday evening with a welcome dinner. It's one of our favorite nights because we get to see so many friends from around the country (and world!). It's like a big family reunion. Thanks to the Internet we can follow our many cystinosis family friends, but nothing beats getting together and catching up.

Our boys, Sam and Lars, didn't waste anytime getting together with their buddies. I had to escort them, along with Henry Sturgis and Jackson Blum-Lang to the lawn so they could be as rowdy as they wanted. It was fun to see all the kids running around in herds.

Friday morning started early with family introductions. A record number of families attended the conference this year! Everyone stood up and shared their wishes for themselves and their children. We wrote our wishes on colorful paper birds and put them on a large paper tree. I wish for the same thing every year – that Sam and Lars will have long, happy and healthy lives. Ashton wished that some day Sam and Lars would be able to be dads.

Nancy started the first session by talking about the many milestones accomplished by the Cystinosis Research Foundation so far. When Nancy finished, I talked about the basics and history of cystinosis – a little primer to provide context for the research talks that followed.

Dr. Paul Grimm talked about the nuances of cysteamine therapy. He talked about both Cystagon® and Procysbi®, and the most effective way to take them. He pointed out the effects of food on the absorption of cysteamine. Many people find if they take cysteamine with food, they have fewer side effects. That's because food interferes with absorption of cysteamine, so it doesn't work as well. Protein and fatty foods are the worst things to take with Cystagon.

Procysbi works best if taken with something acidic, like orange juice. The beads dissolve early if you take it with something with a basic pH, like milk, so you get reduced efficacy. Procysbi should be taken at least two hours after eating, and you should wait at least half an hour after taking it before eating again. Dr. Grimm also noted that the goal is not to get white blood cell (WBC) cystine levels all the way to zero. Carriers of the cystinosis gene (like me) don't have cystine levels of zero. If you use too much cysteamine you run the risk of developing copper deficiency, which leads to collagen abnormalities, skin lesions and poor wound-healing.

Continued on next page

Dr. Mary Leonard, a nephrologist from Stanford, talked about the new CRF-funded study she is doing with Dr. Grimm on bone and muscle health in cystinosis. This is an important topic that has not received a lot of research attention in the past, especially bone health. People with cystinosis have many risk factors for abnormal bone density and structure. Dr. Leonard and Dr. Grimm are planning to do a comprehensive evaluation of 30 children and adults with cystinosis using high-resolution quantitative CT scans, DXA scans and exercise equipment to assess muscle strength. This study will provide background data needed for future studies on possible interventions such as mineral supplementation and hormones. For now, the most effective treatments for good bone health include adequate nutrition, phosphorus and vitamin D supplementation, and weight-bearing activity.

Dr. Bruce Barshop, from the University of California, San Diego (UCSD), talked about a new white blood cell cystine test he has developed. The old WBC cystine test has a lot of variability because there are two main types of white blood cells: granulocytes and lymphocytes. The granulocytes contain the cystine. The ratio of granulocytes and lymphocytes varies from person to person and day to day. During a viral illness, lymphocytes spike to a higher number, so much less cystine is recovered in the lab test, which can give a falsely low cystine level. Dr. Barshop has developed a new test using immunomagnetic beads to separate the granulocytes from the lymphocytes, giving a more pure preparation with more reliable cystine levels. With the new test, individual hospital labs won't have to process the blood anymore. This means any lab can send blood in a yellow top tube overnight to Barshop's lab to be analyzed. This will be a huge blessing for people from small towns or rural areas where hospital labs are not trained to process blood for WBC cystine testing. There will also be a new reference range for target cystine levels. Basically 1.7 will become the new 1.0.

After Dr. Barshop's talk, we had a Q&A panel with representatives from Raptor Pharmaceuticals and Sigma Tau. People were able to voice their concerns about drug access (especially outside the U.S.), cost and insurance coverage, and side effects.

Dr. Sergio Catz, from the Scripps Research Institute in La Jolla, California, talked about his lysosome research. Typically, we think of cystinosis as a disease of cystine accumulation INSIDE the lysosome. However, Dr. Catz has found that because another receptor (LAMP-2A) in the lysosome membrane is impaired, there is also accumulation of proteins OUTSIDE the lysosome, which also leads to cellular dysfunction. He has found a drug that improves cystine emptying from the lysosome by stabilizing a protein called Rab27, which is expressed at lower levels in cystinosis. This work has been done in cell cultures, so the next step is to test the drug in knockout mice.

Dr. Francesco Emma, all the way from Bambino Gesu Children's Hospital in Rome, Italy, talked about screening existing drug libraries for new molecules for treating cystinosis. His lab looked at 1,280 different drugs in cystinosis cells to find molecules that reduce cystine levels and protect the cells from apoptosis (a type of cell death). He found one drug that does both of these things, and it could be a potential new therapy for cystinosis. The good news is that it is already approved by the FDA for another use. His lab is testing it now in knockout mice. Hopefully during the next year he will be able to reveal the identity of this exciting mystery drug!

CRF Board Members Kevin and Teresa Partington

CRF President and Board Chair Nancy Stack opens conference sessions

Board Member Stephen Jenkins, MD, provides an overview of cystinosis for attendees

Next, **Dr. Stéphanie Cherqui,** from UCSD, gave an update on her stem cell research. She explained how hematopoietic stem cell transplantation rescues organ function in mice with cystinosis. She gave us updates on the safety studies that the FDA requires prior to human trials. So far, everything has gone smoothly, and she predicts that the studies will be complete within eight months. She can then go back to the FDA for approval to start the phase I trial. The plan is to recruit two people per year, with a total of six people.

has shown that HSC transplantation can treat an inherited corneal disease.

Dr. Jennifer Simpson, from the University of California, Irvine, spoke about the many ways cystinosis affects the eyes. Typically, we only think about the corneal crystals, but every compartment of the eye is affected, including the retina, conjunctiva, iris and ciliary bodies. One important takeaway was that we shouldn't blame all eye symptoms on the Cystaran™ eye drops. Dry eyes, red eyes or painful eyes can be signs of other eye diseases, such as keratitis and

human trial. The hope is to enroll people in December.

After Dr. Acharya's talk we had the physician/scientist panel. We asked many of the questions people post on Facebook. Several people had questions about muscle wasting and what can be done to stop it. **Dr. Trauner**, a neurologist, noted that there is no effective treatment for muscle wasting, so further research is needed. Levo-carnitine, vitamin D, vitamin B complex and CoEnzyme Q10 are all thought to help muscle function, but there is no hard evidence.

Cystinosis adult panel members share their experiences living with cystinosis

CRF-funded researchers Jennifer Simpson, MD, and Ghanashyam Acharya, PhD

Cystinosis researchers respond to questions from the audience

She has organized the Cystinosis Stem Cell and Gene Therapy Consortium, which is a large group of physicians and scientists who will design the trial and evaluate the participants throughout the study. 2016 is going to be a big year!

After Dr. Cherqui, we heard from **Dr. Celine Rocca**, who works with Dr. Cherqui at UCSD. She presented her research on the effects of HSC transplantation on corneal cystinosis. Her research shows that after allogeneic HSC transplant, cystinosis knockout mice had significant reduction in cystine crystals, restoration of normal corneal thickness, and lower intraocular pressure 12 months later. This is the first time anyone

glaucoma. Dr. Simpson is working on cystinosis guidelines for ophthalmologists, many of whom have little experience treating ocular cystinosis.

Next **Dr. Ghanashyam Acharya**, from Baylor College of Medicine, provided an update on the nanowafer his lab has developed to treat corneal cystinosis. He recently published a paper on the nanowafer technology, which the popular media covered with a great deal of excitement. The nanowafer is so effective that it may replace eye drops for many diseases. Fortunately, Ghanashyam worked with Baylor to give the license for the nanowafer to treat cystinosis to the Cystinosis Research Foundation. CRF is currently working on filing with the FDA to start a

A few people asked about the best time to start eye drops, and whether waiting until the child is symptomatic was too late. Dr. Simpson said children should start the eye drops as soon as they are diagnosed.

Several people also had questions regarding male infertility, since boys with cystinosis develop hypogonadism (a condition in which the body doesn't produce enough testosterone). No one on the panel had much experience in this field, but Dr. Leonard said she would talk with her colleagues in hematology/oncology since they have experience with preserving fertility in young children prior to chemotherapy. We need to recruit a reproductive endocrinologist to the CRF family!

Continued on next page

SAVE THE DATE

April showers bring May flowers
and lots of hope.

Please join us in our **NEW VENUE** for the 2016
CYSTINOSIS RESEARCH FOUNDATION
Day of Hope
FAMILY CONFERENCE
April 7 – April 9, 2016
Island Hotel, Newport Beach, California

"We dreamed of attending the Day of Hope Conference and the reality of was even more wonderful than our dreams.

Once again, you have given us great hope for the future.

The energy and participation was the best ever.

You have taken an already spectacular event to a new level.

The kids all had a blast.

I have never been more optimistic and hopeful about the coming years. Thank you for making that happen."

A happy Jackson Blum-Lang surveys conference festivities

Attendees enjoy time together at lunch on the patio

Paul Grimm, MD, with his son, John, and wife, Kimberly, as he is honored by CRF community members

Shannon Keizer, Katie Ahnen and Natalie Stack at the BBQ

Girls huddle at the BBQ to stay warm

Henry Sturgis in one of many children's activities

CRF Board Member Tom Gendron and Board Vice-Chair Jeff Stack

A family BBQ is the perfect way to end the day

Someone asked about which supplements we need to be careful with. Dr. Grimm noted that taking too much phosphorus at one time can drop the calcium in the blood and cause tetany.

After the panel, **Betty Cabrera**, who works with Dr. Cherqui on the stem cell trial, talked about the Cure Cystinosis International Registry (CCIR). Betty emphasized the importance of registering with CCIR and completing the survey so that scientists will have the baseline information they need to conduct additional cystinosis research. Betty stressed that it's one of the most important things we can do as a community to help find a cure. Even if you've registered before, it's important to update your information annually. CRF has recently revamped the survey to include more pertinent questions for the nanowafer and stem cell trials.

While we were listening to grown-up talks, the kids were back with the babysitters getting royal treatment. They watched movies, played games, did crafts, and ate all the potato chips they could cram in. They received a visit from the "Mad Hatter," a mad scientist, Captain America and a princess named Elsa from a movie I'd never heard of. The kids loved it.

Friday night we had dinner on the beach and lawn. Nancy and Natalie Stack surprised Dr. Grimm with a short film highlighting his life and dedication to pediatric nephrology and patients with cystinosis. They presented him with a special book of photos and letters from his cystinosis patients. There was ice cream, frozen bananas, and those wonderful light-up cotton candy wands. Soon the beach was covered with flashing green, red and blue lights as the kids ran around with their wands.

Unfortunately that was the extent of our participation this year. Our son, Sam, became sick and we returned home to Salt Lake City before the Saturday morning sessions by Drs. Trauner, Ballantyne and Grimm. We also missed the parent-to-parent session, the teen and adult panel, and the fundraising session. Saturday night featured the Natalie's Wish gala with musical guests *Straight No Chaser*. An incredible $2.3 million was raised for research that night from the generous donors in attendance and the many families who raise funds for a cure.

We are grateful for everything CRF is doing to find better treatments and a cure for cystinosis. There is such an incredible community of physicians and scientists who are dedicated to this cause. We were energized by these researchers and knowing that we have so much hope for the future.

We are excited for the upcoming nanowafer and stem cell trials, and the many other things in the pipeline!

Update: *When Sam became ill during the Day of Hope Conference, Stephen and Ashton rushed him back to Salt Lake City with his brother, Lars. They went directly to Primary Children's Hospital, where Sam's nephrologist was on call. Fortunately, the crisis was soon over and Sam is doing much better.*

A Week In The Happy Life of

By Mauricio Vargas, Antonia's dad, Bogota, Colombia

We are the

"Mauricio + Marcela =

from Bogota, Colombia. I work in marketing and Marcela is dedicated to taking care of our daughter. Our main challenge is to give Antonia a happy life. That's why I wanted to introduce her to you and give you a brief description of a typical week in her life.

Antonia

Antonia has a happy life, which is what we wanted to share with you. Of course, we have the same issues as other cystinosis families. Every two months Antonia has a blood test, which she calls her "Brave Girl Mission;" she visits her doctors, whom she loves; her first word was "water;" she is not the tallest girl at school; she has difficulty swallowing food; the G-tube is part of our life; the nutrition pump and medicines are our luggage; and we pray daily for better treatments and the cure. This part of the story you know very well.

We wanted to introduce Antonia to the Cystinosis Research Foundation as a representative of the 20 cystinosis cases in Colombia. In Columbia, we have two challenges: first, a lack of knowledge about the treatment for eyes since we have no access to cysteamine drops, and our most important challenge: no laboratory offers the cystine level test. By working with CRF, Marcella and I are confident that we can find ways to solve these problems for cystinosis

Antonia team" (M+M=A)

patients not only in Colombia, but for others throughout Latin America, who have additional issues, which I will discuss in future articles.

"Why us?" It's a question we've never answered. We understand that Antonia chose us as her parents because she saw in us the ability to fight on her behalf and meet the challenge of giving her a HAPPY LIFE.

Dedicated to our Army of Angels: Nancy Stack, who made our trip to the CRF Day of Hope possible; Dr. Luz Stella Gonzalez; Dr. Nena Lung; Dr. Felipe Ordoñe; Dr. Hernan Robles; Dr. Mauricio Lozano; Dr. Ricardo Gastelbondo; Dr. Hernando Silva; Roland Kalt; Dr. Leticia Belmont; Dr. Maria Helena Vaisbich; the Sigler-Magnus Family; and Denice Flerchinger.

MONDAY

Every morning Antonia uses a towel on hair to pretend that she is a princess. Rapunzel is one of her favorite characters. Antonia arrives at the German school she attends at 9 a.m.

She is known for her independence, creativity, intelligence, tenderness, ability to speak, and her love of music and drawing. During the week, she takes chocolate milk and natural juice in the morning, and she eats soup, rice, vegetables and meat for lunch. Yes, you guessed – in small quantities. At 2 p.m. she's back at home, where she watches TV, plays with her iPad and paints different circles, which she says are princesses. Finally, at 7 p.m. she goes to bed.

TUESDAY

The routine is almost the same as the previous day. The only difference is that Antonia attends ballet class. When she returns home, she gives us lessons, but first we need to warm up like "Angelina Ballerina."

WEDNESDAY

She wakes up happy because today is swimming day. At 10 a.m., Antonia, who is wearing her swimsuit, jumps into the pool and, as if by magic, becomes the "Little Mermaid." Once home, she is very tired, so it's time for rest.

THURSDAY

After her classes, Antonia attends soccer practice. Our greatest rewards are the goals she scores and dedicates to us each Wednesday.

FRIDAY

"Family time" starts with a movie or dinner in town. In restaurants, she asks to see the menu and often chooses chicken soup. She usually sleeps in the car on the way home, but she wakes when we get home and stays up late fulfilling her dream of singing like Anna, Elsa or Olaf from *Frozen*.

SATURDAY

Every Saturday she attends riding lessons. Her horse is named "Mexico." She loves talking to him but when we ask her what Mexico said, she answers, "Animals can't talk." After the class, we enjoy the sunny days in Bogata.

SUNDAY

It's time to go to church. We have a lot to be thankful for.

Dear Nancy and Jeff,

From the moment Day of Hope ended, I wanted to write what I was feeling, but I preferred to come home and do it calmly.

I want to start by telling you **THANK YOU…**

THANK YOU for opening our doors to hope. Because of CRF we believe and have faith that soon our are most coveted dream – the cure – will come true.

THANK YOU for making Natalie's wish a reality. Now, it is also our wish.

THANK YOU for being so special, for your warm welcome, and for making us feel at home even though we were thousands of miles from our home in Bogata, Colombia.

THANK YOU for your words, "Your children are my children," which made us feel welcome in this great family.

THANK YOU for giving us a big hug, which gives us tranquility, support and complicity … we speak the same language.

THANK YOU for showing us that there are no limits to what we can achieve – this belief is a great motivation for the Quijano Vargas family.

THANK YOU God for life and for putting CRF in our way.

THANK YOU for bringing together the best medical and research team. We ask God to continue to inspire all the doctors, so they will continue their work for our children.

THANK YOU for inviting us to be part of this beautiful and strong group of families.

From Colombia, we only have immense **GRATITUDE**. We are sure that life will continue to bless your family.

We look forward to attending next year's Day of Hope. All the progress, news, union and love nourishes our souls.

Warm hugs,

"Mauricio + Marcela = Antonia team" (M+M=A)

CCIR & YOU

PROPELLING CLINICAL TRIALS FORWARD

Imagine a world where a child born with cystinosis can be effectively treated so that kidney complications such as Fanconi syndrome and kidney transplants are avoided altogether.

Imagine better, more effective drugs that prevent problems with eyesight and muscle wasting and that have fewer side effects.

Dedicated researchers are working tirelessly to bring such a world within reach using innovative research methods and cutting-edge technologies.

Propelling laboratory research all the way forward to human clinical trials, however, will require help from the patient community. There are many milestones along the way to drug development that simply cannot be reached without patient support and active participation.

Imagine that a potential therapy reaches the clinical trial phase but not enough eligible study participants can be identified to take part in the study.

Alternatively, imagine that clinical trials that establish safety and efficacy of a drug are successfully completed, but that insufficient information about cystinosis is available for the Food and Drug Administration to evaluate whether there is a pressing need for the new therapy on the market. These obstacles to drug development for rare diseases are common and can cause significantly delays if we are unprepared to deal with them.

Luckily, the cystinosis community can overcome or avoid these obstacles by registering with CCIR today and by keeping their CCIR profiles up to date. CCIR is an online patient registry where medical surveys that patients and their families complete are confidentially stored. The anonymous data is made available for purposes that will advance medical research in cystinosis. However, the registry is so much more than a database. It is a tool meant to empower the cystinosis community. Participation in CCIR is a way for you to advocate for yourself or on the behalf of loved ones that improved therapies are needed.

Without your active participation, CCIR has no real value. We need you to participate and update regularly. We also need you to encourage others to register. By banding together, we can reach those better places that we can now only reach in our imaginations.

CCIR CURE CYSTINOSIS INTERNATIONAL REGISTRY

www.cystinosisregistry.org

IMPORTANT CHANGES TO THE SURVEY

The CCIR survey has been revised to better capture what it is like to live with cystinosis and to understand how currently available treatments address patients' needs and concerns. Several cystinosis experts were consulted to improve the survey so that gaps in our knowledge about cystinosis could be addressed.

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

Currently registered patients will receive an email announcing that the improved survey is available. Please visit the CCIR website now so we can begin to collect this important information.

WHAT TO EXPECT

You will notice that 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 you response.

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

Betty L. Cabrera, CCIR Curator

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

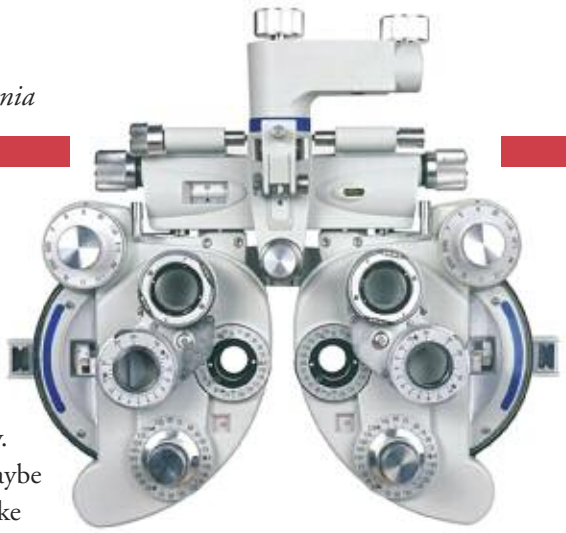
Seeing a HEALTHIER Future

Dear Jenna and Patrick,

On Thursday, you will go to your ophthalmologist for an appointment that dad and I have been dreading since the day you were diagnosed with cystinosis. It's not because eye doctor appointments are scary or painful, or because you have bad eyesight. We simply and sadly realize that in spite of your stellar compliance to your daily medications, the crystalized cells in your body caused by cystinosis have accumulated over the years. These cysteine crystals have really begun to build up on your corneas.

After hearing Dr. Simpson speak at the Day of Hope Conference, we realized that you must immediately begin the eye drop treatment that is required to maintain your vision.

By Teresa Partington, Jenna and Patrick's mom, Sacramento, California



It is important to us that you both understand that your dad and I are really sorry you have to take so many medications. We are especially sorry that you now have to add another one to your daily regimen. Like all your other medications, you cannot miss doses of the eye drops, or your cystinosis will progress and make you sick. You must put the Cystaran™ drops in your eyes every hour, every day. I know it will be very hard at first. We may all get frustrated, and maybe even sad. But I pray that it will eventually become second nature. Like drinking lots of water, it will eventually become part of your day.

Lots of people use eye drops for lots of different reasons. The eye drops are safe and your eyes will get used to them. Jenna, after a few months you will be able to go outside without sunglasses. Patrick, you haven't developed a desperate need for sunglasses yet. We think starting the drops now will keep you from experiencing the pain Jenna feels when she is in bright light.

Jenna, do you know why the sunlight hurts your eyes so much? Well, the cystine crystals on your corneas are like a thin film of diamonds or glass prisms. When sunshine or any bright light hits a prism, the light is multiplied and reflected in a million directions. If there are a million crystals on your eyes, all reflecting light in a million directions, it is going to be painfully bright!

Your eye doctor will prescribe the eye drops at your appointment this week. They are a specialty drug that was approved by the FDA just a few years ago. We are very lucky to have insurance that allows us to get the eye drops and all of your other medications. If you use the drops properly, they will dissolve most of the crystals on your corneas and help keep your eyes healthy.

We don't want to scare you, but there are children with cystinosis who have never had these eye drops – or who don't follow the rules for taking them. Many of those children cannot go outside or turn the lights on because the brightness causes so much pain. Some of those children have even gone blind. The cystine builds up gradually, even when you feel fine. The crystals are always forming, so you need to be diligent about taking your medications and using your eye drops.

Jenna and Patrick, you are both amazing kids. You are so good about managing your burdensome health challenges. You are happy and cheerful, and never complain. We are proud of you and we love you just the way you are. You are special and unique – and you teach your dad and me, and everyone in your life, so much about being brave and strong and determined.

Your dad and I hope you always remember how lucky you are in spite of how hard things may seem at times. It's fine to have a down day or two occasionally as long as you ultimately remember your blessings and perk back up and live happily. Let's plan on some rough days while we all get used to the eye drops, okay?

We know you will always take good care of yourselves. Your dad and I are here to help you and support you. We want to teach you and guide you to become independent and able to manage your health on your own. Can we keep working together on that?

With lots and lots of love,
Mom and Dad

An Awesome Day with One of the Guys

By Kristen Bruce,
a Partington family friend,
Sacramento, California

As a parent of a child without cystinosis, I can tell you that coming to the *Day of Hope Conference* was both humbling and inspiring. Humbling, as I learned even more than I already knew about the constant challenges that a cystinosis family deals with every day. Inspiring, because of the amazing progress CRF has made, as well as the love and support I felt among the families. What a beautiful community of people!

I am so grateful that Teresa and Kevin have the wonderful CRF community walking with them on this journey. You are all amazing.

Living just two doors away from the Partingtons, Jenna and Patrick have become a part of our daily lives. As far as my 10-year-old son Nick is concerned, Jenna and Patrick are simply two regular pals on the block! While Nick knows that his buddy Patrick takes medicine for his kidneys, he has no idea of how hard Patrick's body is working to fight cystinosis every day.

By bringing Nick to the *Day of Hope Conference,* I thought he might gain a better understanding of cystinosis. While playing in the kids' club, I thought he would recognize the possible similarities among the kids with cystinosis – such as wearing of sunglasses, fair complexion and light hair. But that was not the case. What I learned was that for Nick, the trip was no more than an "awesome day" to play and meet some of Patrick's friends. And that was that – a true 10-year-old's perspective.

I struggle with wanting Nick to understand the seriousness of cystinosis. If he understood more about what Patrick endures (without one complaint to his pals at school), he would surely see that Patrick is the strongest kid he knows!

I know he would gain a whole new admiration and respect for his pal. But for now, that will have to wait. Patrick is simply "one of the guys." Cystinosis won't get in the way of that.

While Nick may not understand what the *Day of Hope Conference* provided for so many, I certainly do. I am so grateful that Teresa and Kevin have the wonderful CRF community walking with them on this journey. You are all amazing.

Thank you for welcoming us. I am so glad my son and I could be a part of such a special weekend.

From left, Nick Bruce, Patrick Partington, Hadley Alexander, Elise Thomas and Jenna Partington at the 2015 Day of Hope Conference.

By Dinny Thomas,
a longtime Partington family friend,
Sacramento, California

Dinny Thomas with Teresa Partington one hour before Jenna and Patrick's birth, on December 7, 2004

IT'S GOING TO BE OKAY

It's rare to meet someone as a young adult and know that person will be in your life forever. That's what happened to me when I met Teresa. It was spring of 1998 and our paths crossed because the boys we were dating, Ron and Kevin, were roommates and business partners.

Those were undoubtedly some of my most treasured years. Trips, dinners, game nights… the four of us were inseparable. It was a natural progression when we were engaged within six months of one another, married within a year of each other, and then starting the journey of growing our families.

I was pregnant first and had my first child, Ayden, in the summer of 2003. Naturally, my friend was there every step of the way. Teresa helped soothe my fears and concerns, as well as celebrated with me the anticipation and joy of being a new parent.

Then, it was her turn to be pregnant – because that's just how things always were with us. We seemed to live parallel lives and our friendship only deepened because of it. When she shared her own fears and joys during her pregnancy, I told her, "It's going to be okay."

One night, Teresa and Kevin took Ron and me to Paragary's for dinner after work and shared their ultra-sound picture with us. We studied the photo closely and realized they would be doubly blessed with twins! I was lucky to be in the room when Jenna and Patrick came into this world.

Continued on next page

What a glorious day it was! Not knowing what sex the babies were, I think I almost passed out from holding my breath in anticipation of seeing who they would be! First Patrick, and then Jenna. They were perfect.

Now, Teresa and I were new mommies together, and having her two streets down in our Land Park neighborhood was so much fun. Walks to the park, play dates, and long days were made all that much better with my friend by my side. I remember one day Teresa asked me, "Does Ayden stay in his high chair for over an hour when he eats?" To be honest, I was jealous because my son wasn't sitting anywhere for more than 30 seconds!

Then came her babies' water consumption, coupled with horrific diaper rashes. I'm certain we tried every diaper rash cream on the market. Teresa then called me and asked if I could run over to see if their breath smelled fruity? She was concerned that they may have diabetes. We were both scared – something wasn't right. But I told her, "It's going to be okay."

Jenna, Ayden, Elise and Patrick play in Lake Tahoe. Summer 2013

Soon after their one-year check ups, and after a frightful hospital stay for Jenna, my friend's babies were diagnosed with a rare genetic disease called cystinosis. I will never forget when I first typed the word c-y-s-t-i-n-o-s-i-s into the computer search engine. The words on the screen were unreadable because I couldn't fight back the tears. Again, I reiterated those five little words, "It's going to be okay," to my friend, but this time I wasn't entirely sure.

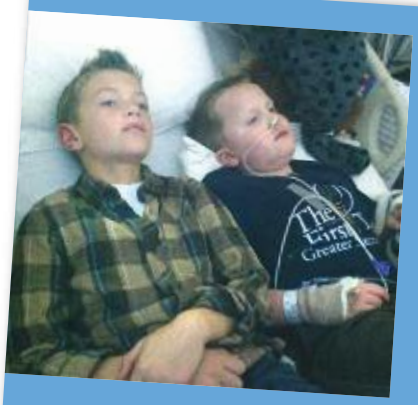
And so… our group of friends went into fix-it-mode. We started Jenna & Patrick's Foundation of Hope and organized our first walk and our first golf tournament. Then, Teresa and Kevin met Jeff, Nancy, Alex and Natalie Stack, and the rest is history.

Over the years (and especially while writing this letter for *Cystinosis Magazine),* I have thought a lot about those days that Teresa and I shared together through our courtships, our pregnancies and the births of our children.

I have relived the pain of discovering that Teresa's twins – my Godchildren – had a very rare, incurable disease. As difficult as it has been, this amazing family's journey has been a privilege to witness and to be a part of. Their strength and their pain, their joy and their love has taught me the importance of hope in my own life experiences. I have learned that so often, friendship is what carries us through life's happiest and darkest days.

Our community has an abundance of love for the Partington Family, and I have watched them contribute generously to the cystinosis cause over the years. Next spring, Ron and I will participate in the annual *Swing & Bling* golf tournament and dinner event to benefit Jenna & Patrick's Foundation of Hope. The funds raised will be passed along to the Cystinosis Research Foundation at the Natalie's Wish Gala, which I have been privileged to attend over the years. I am grateful my friend has the remarkable CRF organization to guide her and her family in this often difficult, and ultimately hope-filled journey.

This year, my daughter Elise and I were able to attend the Day of Hope Family Conference. It was humbling and encouraging to be a part of this gathering of families, doctors and researchers…all working toward the same goal: A cure for cystinosis. I could see that, because of this annual gathering, families are able to move from a place of worry and fear to a place of hope and empowerment.



A LIFETIME OF LOVING FRIENDSHIP AND SUPPORT

Above: Ayden Thomas rests with Patrick after a medical procedure

Top right: Ron and Dinny Thomas, 1998

Right: Jenna and Elise Thomas at this year's Swing golf tournament, March 2015

And so… this story of friendship will go on. And I know, especially after attending this year's Day of Hope Conference, I can be confident when I tell Teresa, "It's Going to Be Okay."

SWING5

Jenna & Patrick's Foundation of Hope

Thank you to our State Street volunteers!

Ron and Ayden Thomas on the tandem bike that was raffled off.

This year's *Swing* golf event on Friday, March 27, benefitting Jenna & Patrick's Foundation of Hope, was a spectacular success!

The move this year to the exclusive Catta Verdera Golf Club in Lincoln, California, and the beautiful weather contributed to a fantastic day of golf for a great cause. This year's sold out tournament with 132 golfers raised more than $110,000 for cystinosis research. Thanks to our generous sponsors and donors we are growing closer than ever to a cure for cystinosis.

Congratulations to the winning foursome from Sacramento's Heller Pacific Group. The winners will enjoy the grand prize trip to Bandon Dunes Golf Resort next October!

All funds raised by Jenna & Patrick's Foundation of Hope are sent to the Cystinosis Research Foundation. CRF's Scientific Review Board, comprised of leading cystinosis scientists from around the world, reviews research applications and recommends who should receive a grant.

SAVE THE DATES

Mark your calendars for next year's Swing & Bling event: Thursday, March 10, golf at Catta Verdera Golf Club and Friday, March 11, gala dinner at The Citizen Hotel. For updates, visit www.jennaandpatrick.org.

Jenna & Patrick's Foundation of Hope

CALEB'S CAUSE

A CRF PARTNER FOR A CURE

On Saturday, January 24, 2015, Caleb Gowan's family and friends gathered for *Caleb's Cause* – a spaghetti feed and silent auction to raise money for cystinosis research. Caleb's cousin, Brian Gowan, led the event by organizing both the supper and auction, along with a $5,000 cash drawing.

Many volunteers, including Caleb's 4H group, helped with the event. Local businesses graciously donated items including bicycles, pieces of art, an autographed hockey jersey from Matt Green of the Los Angeles Kings, jewelry, gift certificates and many other items.

Caleb's Cause raised more than $26,000 for cystinosis research. Caleb and his family are still overwhelmed by the generosity of local communities that came together for such a great cause!

By Jana Riley-Kraulik,
Caleb's mom, Karlstad, Minnesota

UPDATE

CALEB'S TRANSPLANT

We sure didn't expect things to happen so fast. Caleb's doctors were planning to put a port in him to start dialysis. Fortunately, after his very sudden kidney transplant surgery early on April 25, Caleb is doing great and he won't need dialysis.

So far his body is accepting the kidney and overall he is able to tolerate the antirejection medications. He's such a fighter and did very well in the hospital. I'm very proud of him.

Since leaving the hospital, a week after the transplant, Caleb has only taken two Tylenols for pain and none of the oxycodone that was prescribed to him.

His energy levels are great and on Mother's Day, two weeks post-transplant, he was shooting baskets at a park next to our hotel. I am amazed by his progress.

God had the perfect timing for his transplant. The week before his surgery, we were able to spend time with our cystinosis family at the Day of Hope Conference in California. He had the transplant so quickly that we had little time to worry about it before hand.

Please pray that Caleb's body continues to accept this kidney and for the family who lost their loved one. It was someone around the age of 45 who probably had a lot of life to live.

Finally, thank you to everyone who signs up to be an organ donor on their driver's license! What an impact their choice has made in our lives!

To learn more about donating a kidney, visit www.kidney.org.

JUST ISN'T ENOUGH FOR HANK

It was a crazy winter in North Idaho. Lack of snow forced us to cancel our 7th annual fundraiser. 24 Hours of Schweitzer had been scheduled for March, luckily we had a backup plan ...

In August of last year Hank's 70-year-old Grandpa, Dave Sturgis, committed to racing his bicycle in Race Across America (RAAM), a grueling non-stop 24-hour-a-day team-race across 12 states. RAAM is widely recognized as one of the world's toughest endurance bicycle races and is 30 percent longer than the Tour de France. The race begins in Oceanside, California on June 20, 2015, and doesn't stop until the finish line in Annapolis, Maryland. Dave is part of Team Laughing Dog, a four-person team from Sandpoint, Idaho. The team's goal is to raise money and awareness for 24 Hours for Hank, a non-profit organization dedicated to finding a cure for cystinosis.

Dave began training for the ride in October 2014 and is currently training six days a week. Team Laughing Dog's goal is to complete the race in seven days.

Dave says, "I'm doing something this hard because I know every single day is hard for Hank. I promised my grandson that I would help to make his disease go away forever. Raising money to support cystinosis research will help ensure Hank's future by finding a cure for this deadly disease."

As the word spread about the cancelation of the ski event and Dave's promise to Henry, teams that usually participate in the ski event redirected all of their fundraising efforts to help Dave reach his goal.

The team had already started to plan some fundraisers, and on April 24 we held our first ever golf tournament at Avondale Golf Course in Hayden Lake. It was a great success and made a large contribution to the fundraising goal.

On July 2, three days after returning from the finish in Annapolis, we will be having a dinner and auction keynoted by Ben Stein in Sandpoint, Idaho. Having these two new fundraisers has introduced 24 Hours for Hank and cystinosis to a whole new group of people. About 75 percent of people attending these events have never heard of our organization or cystinosis.

2015 has been a big year for Henry! He is now eight years old and in second grade, and he is taking all meds orally. During spring break he even had his G-tube removed. We had given him a goal of six months without relying on it and in April he reached that goal. Henry continues with occupational and physical therapy weekly. And he's excited that summer is about to begin!

RACE FOR A CURE

My name is Dave Sturgis and Hank is my grandson. Henry has a rare metabolic disease called cystinosis. Cystinosis is an incurable disease that affects approximately 500 people, mostly children in the United States, and fewer than 2,000 worldwide. Because of the very small patient population, cystinosis research is primarily funded through private donations.

Henry was diagnosed in December 2007, when he was 16 months old. Today, he takes 43 doses of medicine each day to maintain his condition, and hopefully stabilize the disease. He also takes hourly eye drops to help prevent blindness, and a growth hormone shot six days a week.

I recently turned 70 and am riding in Race Across America, a grueling coast-to-coast ride that begins in Oceanside, California, and finishes 3,000 miles later in Annapolis, Maryland. I am participating in what is called "the world's toughest bicycle race" to raise money to help find a cure for cystinosis.

In June of 2015 I will be on my bike, racing across America – but the most important race is to find a cure for cystinosis for my grandson Henry and all those who live with this dreaded disease. To date, my Laughing Dog Team mates and I have raised $56,000 of our $100,000 goal. With your help, we can reach, or even exceed our goal, and make a life free of cystinosis a reality. But time is of the essence.

To donate visit: 24hoursforhank.org.

Tee'd Up for a Cure

On April 24, 112 people came to play golf and raise money for cystinosis research at Avondale Golf Course in Hayden Lake, Idaho. At the end of the day more than $18,000 was raised for research!

The scramble included longest drive, closest to the pin and a putting contest. Additional games – including winning a car for a hole in one, but no one was lucky enough this year – thrown in to keep the day interesting. Longtime supporter Team Hank E. Panky sold festive jello shots on the turn to keep everyone going on "the back 9." The fun-filled tournament was followed by a delicious prime rib dinner.

Having a first-ever golf tournament allowed us to meet a whole new group of supporters, many whom had never heard of 24 Hours for Hank or cystinosis. We sincerely thank everyone who played or volunteered, making the day such a tremendous success.

Join us on Thursday, July 2, 2015 for our *DINNER WITH BEN STEIN,* at the beautiful Trinity at City Beach, Idaho. Ben is an actor, author, columnist and longtime 24 Hours for Hank supporter. For tickets: tricia.sturgis@gmail.com

DO YOU HAVE A ROLE MODEL?

I want to tell you a story about mine...

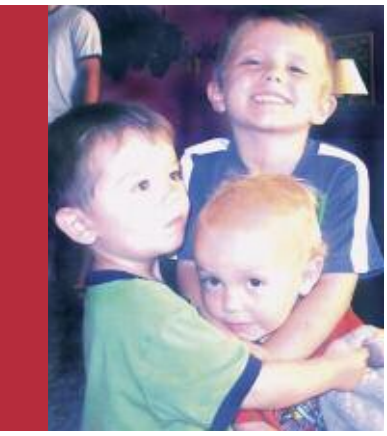
By Rob Timmons, board member, 24 Hours for Hank

"A role model is a person whose behavior, example, or success is or can be emulated by others, especially by younger people."

I am incredibly fortunate to have known the Sturgis family for the past 30+ years and during that time they have treated me as part of their family. I can honestly say that their guidance, support and life lessons are those that I try to teach to my boys every day!

I could literally write a book (maybe I will someday) about the lessons I have learned from Dave Sturgis and the one-liners I have heard from him. Now, my boys have heard every one of them as well! One lesson has always stood out to me. Dave again reminded me of it, in November of 2007, after my best friend's son was diagnosed with cystinosis:

"Lots of people talk about doing something, very few take action."



In May of 2008, with the support of some of the most amazing people I have ever met, we took action and started our journey to find a cure for cystinosis.

This past summer, on the last night of their vacation, I again raided the Sturgis family gathering in Priest Lake, Idaho. While we were sitting around the fire pit, Big Dave told me about an opportunity that he had been given to help raise awareness for cystinosis. He was going to ride his bike across America as part of a team that has been supporting cystinosis since the beginning.

That conversation was short and to the point as I reminded Dave of the lesson he taught me many years before:

"Lots of people talk about doing something, very few take action."

Big Dave is now 70 years old and has spent every day for the last 11 months training on his bike. He and his amazing teammates will start their journey on Father's Day. They will ride more than 3,000 miles for one reason: to help raise awareness and money to find a cure for cystinosis!

I am sure everyone reading this has a role model. My advice is to make sure they know the impact they have had on your life and to tell them thank you!

Help me say thank you to Big Dave and Team Laughing Dog, by donating to their ride at www.active.com/donate/24hoursforhank2015/raam.

From Governor Otter

The First Lady and I would like to share an inspiring story of two Idahoans that underscore some of the great qualities of our citizens. Hank Sturgis is a young man courageously fighting a particularly cruel and rare disease. He has cystinosis, which affects 500 children around the nation, and about 2,000 worldwide.

Cystinosis is incurable and slowly destroys every organ in the body, including the kidneys, eyes, liver and brain. Because it is so rare, research money for cystinosis is scarce.

However, that is not stopping Hank from fighting it or his grandfather from traveling across this country in an effort to help find a cure.

Dave Sturgis is participating in a 3,000-mile bike race to raise money and awareness. At the age of 70 he decided to enter the Race Across America that starts in Oceanside, California on June 20, and ends in Annapolis, Maryland, later this summer. Dave is part of a four-person team from Sandpoint that will compete in the event, which they hope to complete in seven days.

We admire the selfless determination that Dave is using to propel himself and his team across thousands of miles and bring his grandson Hank and others with cystinosis closer to a cure.

While Dave's effort is aimed at raising awareness, it's also a moving expression of love for his grandson and hundreds of other Americans. Lori and I will be following his progress on the Race Across America and in the race to beat cystinosis.

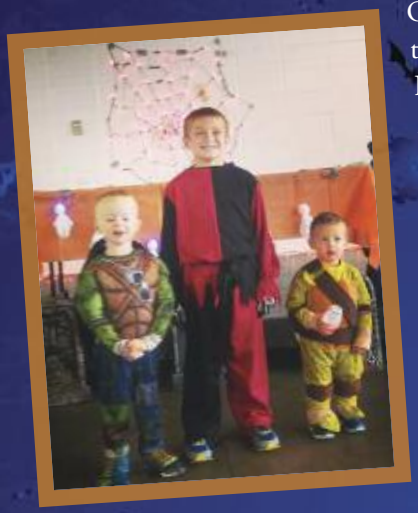
If you would like to help Dave, visit www.24hoursforhank.org

Or send donations to: 24 Hours for Hank P.O. Box 2564, Sandpoint, Idaho 83864

IDAHO GOVERNOR C.L. "BUTCH" OTTER

Lots of Love for Landon
A Halloween Gathering

On Saturday, November 1, 2014 the Hartz family held its 4th Annual *Lots of Love for Landon Halloween Gathering.* A record-breaking 366 guests attended the celebration to honor Landon Hartz. Friends and family of the Hartz family came and – to help swell the size of the crowd and grow the proceeds – they in turn brought their own friends and family. The response from the community in which Landon and his family lives was absolutely amazing.



Chandler Moore (center), who also has cystinosis, made friends with Landon and his little brother Jordan.

One hundred guests under the age of 12 created an atmosphere that was incredibly lively – some might say "spooky" – to say the least. Best of all, Landon just loved running around with his buddies from school, his play group and his neighborhood.

Some great family members and friends planned the event, set it up, ran the festivities and cleaned up when it was all over. The event simply couldn't have happened without these passionate, generous, hardworking people.

Guests were entertained by DJ Mark "Hypnotyza" Raich, who volunteered for the third time. Mark didn't know the Hartz family prior to volunteering his services the first year; fortunately he has made a huge difference every year since then.

Adults, kids, couples and groups paraded around the room to show off their costumes to the large crowd. Winners were selected in five categories, including best boy costume, best girl costume, best adult female, best adult male and best couple/group costume. Costumes get better – and crazier – every year, which makes choosing winners that much tougher.

Top: Jimmy Hartz with his cousin Brenda Brown, whose employer generously donated signs and printed materials for the event.

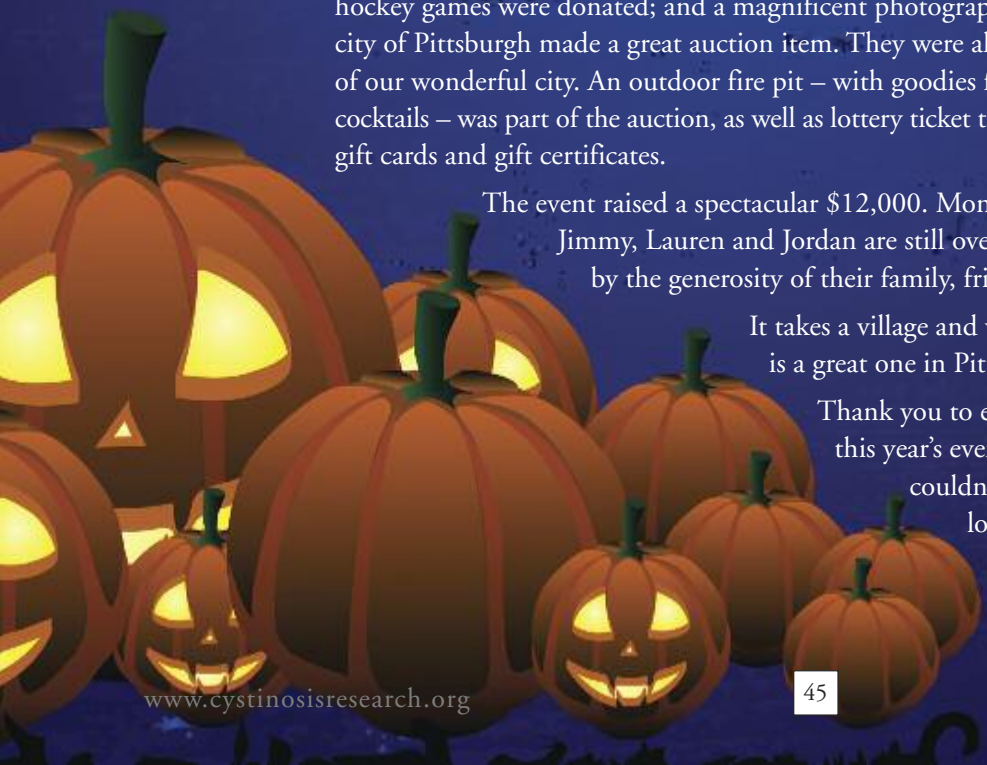
Below: Lauren Hartz, Landon's mom, and DJ Mark "Hypnotyza" Raich presented prizes to the lucky winners.

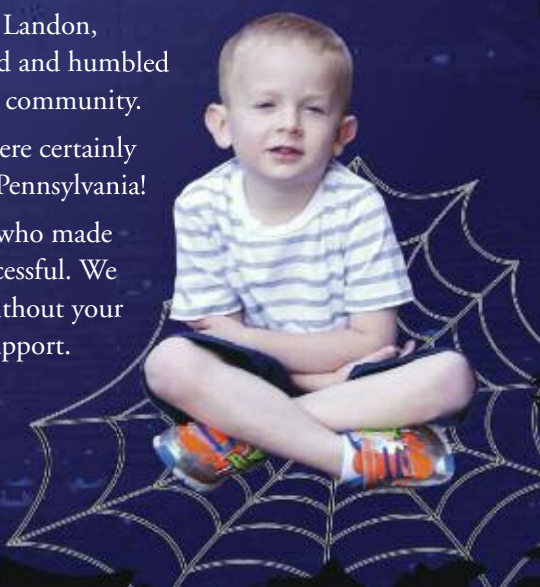
More than 30 Chinese Auction prizes were presented and 20 door prizes were raffled off. Landon's family and friends worked hard to seek out and make baskets for the Chinese Auction. Handmade corn hole boxes were built and painted with love; tickets for Steeler football games and Penguins hockey games were donated; and a magnificent photograph of the beautiful city of Pittsburgh made a great auction item. They were all representations of our wonderful city. An outdoor fire pit – with goodies for s'mores and cocktails – was part of the auction, as well as lottery ticket trees, and countless gift cards and gift certificates.

The event raised a spectacular $12,000. Months later, Landon, Jimmy, Lauren and Jordan are still overwhelmed and humbled by the generosity of their family, friends and community.

It takes a village and wow – there certainly is a great one in Pittsburgh, Pennsylvania!

Thank you to everyone who made this year's event so successful. We couldn't do it without your love and support.





By Jessica DeDio, Bailey's mom, Fullerton, California

3rd Annual

Bailey Believes

HIS HOPES, HIS DREAMS AND A CURE

Dirt Bike Ride for a Cure

Bailey with his friends, Dustin (left) and Josh (center)

Jason Brown with Bailey. Jason organizes Bailey's Dirt Bike Ride each year

On November 8, 2014 we had our 3rd Annual *Dirt Bike Ride for A Cure* in Barstow, California. It was a beautiful Saturday morning in the desert and another successful year filled with love, friends, camping and dirt bike riding.

Thanks to all our Bailey Believers and wonderful support community we raised $22,320 during this Ride – and we had a great time doing it! I am so very honored and touched by all the love people have for my amazing son, Bailey, and his friends who suffer from cystinosis.

SAVE THE DATE!

Our 4th Annual *Dirt Bike Ride for a Cure* takes place on Saturday, November 7, 2015.

If you can't join us at one of our Bailey Believes events, please consider donating online at: www.cystinosisresearch.org/donate-for-bailey

Mary Jordan, Jessica DeDio and Bonnie Paju

We had about 50 riders in the 13-mile ceremonial loop with Bailey and lots of supporters just camping out for the weekend. This year was extra special, because this was Bailey's first ride since receiving his new kidney from our hero Mary Jordan, who was also there to watch Bailey tear it up on his dirt bike!

We were very proud to have a Donate Life booth on display, thanks to our friend and Donate Life ambassador Jennifer Barnette. Along with raffles and auctions, we enjoyed great music during the day and karaoke by bonfire at night, thanks again to our friends Michelle and Stacey Hayden.

Every year, our hearts are overwhelmed with love, hope and gratefulness. I want to thank each of you for coming out, and loving and supporting Bailey and our family, and choosing to help us find a cure for cystinosis. Your thoughts, prayers and unconditional support do not go unrecognized. Thank you for giving us something to believe in.

As I write, my beloved friend, Shannon Paju, is in declining health from cystinosis. Shannon was unable to attend our recent Dirt Bike Ride for A Cure. However, during our second Ride, she rode in a Razor for the first time ever with my mom, Sherry Boucher. After that Ride, Shannon told me it was the best day in her entire life. I am praying for Shannon and all those who are battling with cystinosis for more best days in their lives.

Love always, always believe,
Jessica DeDio

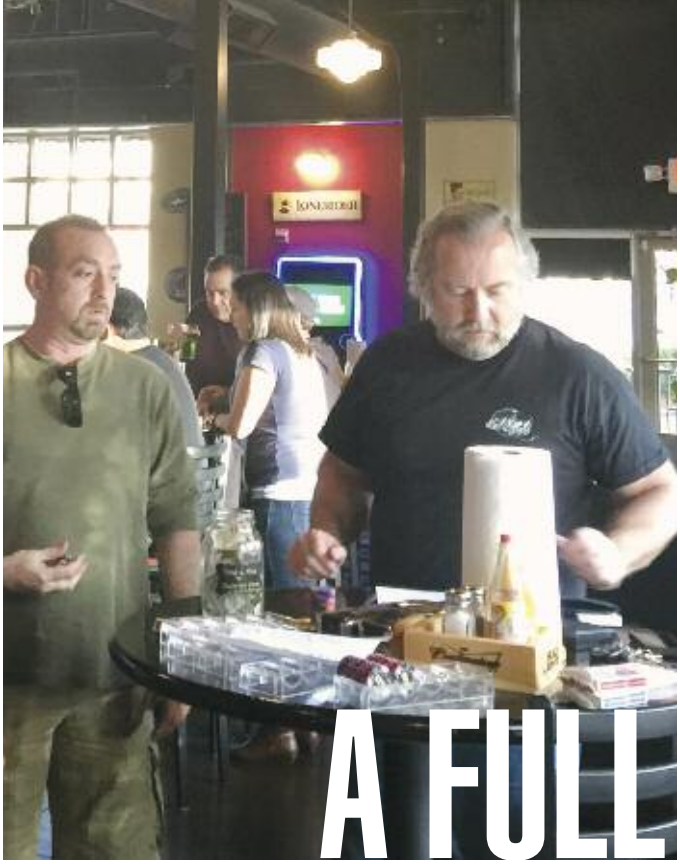
Bailey's Doing Great

Bailey's been doing great since his second chance at life! He likes to be outside all the time. He's either in his Man Cave (shed) working on his motorized bike, building something, or riding around town with his friends. He will be 18 years old this November and after finishing high school he's considering attending school for Auto Mechanics.

In October 2014, Bailey donated blood at UCSD to Dr. Stéphanie Cherqui's study for gene modification of human stem cells in cystinosis patients. This is the third study he's participated in for cystinosis research.

On April 25, 2015, Bailey and a team of Believers participated in the 5K Donate Life Walk for the second year in a row. Unlike last year when Bailey was walking much slower – it was only three months after his kidney transplant – this year he stayed ahead of our whole team throughout the entire walk!

Now, we're looking forward to the 4th Annual Dirt Bike Ride for a Cure on Saturday, November 7, 2015. For information about our upcoming events visit www.facebook.com/BaileyBelievesDonateLife



Kurt Payne and Tom Wiegand (right)

Keegan Manz (right) with his older brother Shane and his mother Nicole

A FULL HOUSE AND A BIG HEART FOR CYSTINOSIS

By Rebecca Hite, Tom Wiegand's wife

"Some people spend an entire lifetime wondering if they made a difference in the world. But, the Marines don't have that problem." RONALD REAGAN, PRESIDENT OF THE UNITED STATES

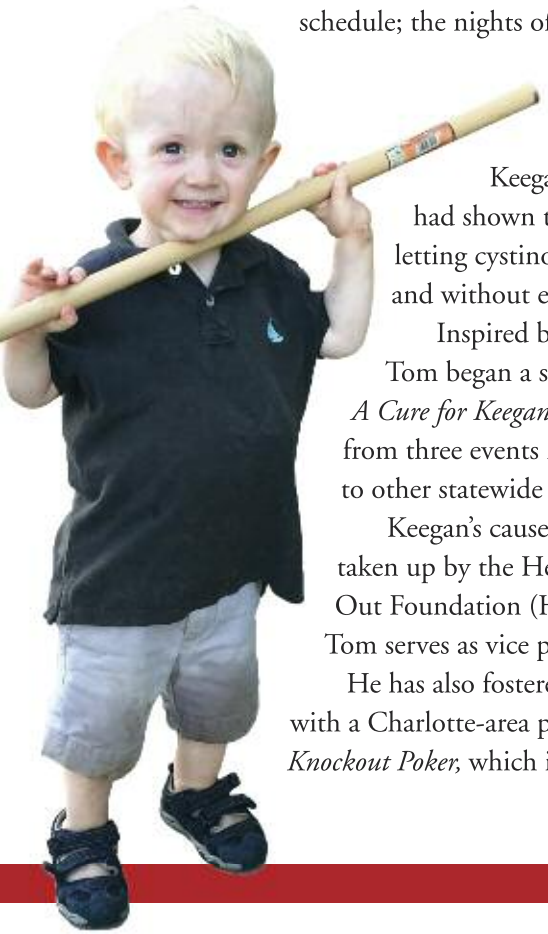
Tom cannot hide the fact he was and still is a Marine. Just by looking at him and speaking to him he epitomizes the U.S. Marine Corps. At 17, Tom enlisted in the Marine Corps because he wanted to serve his country. He enjoyed "every minute" of being a leatherneck, retiring after a success-filled 20-year career.

Today, Tom continues the Marine Corps mission of taking an active role in safe-guarding the lives of children via civic engagement. Tom organizes the Marine Corps Toys for Tots drive each Christmas, as well as works with the Big Brothers program. In 2011, Tom was able to deliver three truckloads of toys to children and families in need in the Raleigh-Apex area.

A lifelong poker player, Tom formed his own Raleigh-area poker league. In 2015, he incorporated his company under the name Knockout Poker USA. Tom's company is dedicated to developing patrons' poker skills, while fostering a new generation of poker professionals. He specifically selected poker because he believed it provided opportunities for community members from all backgrounds to engage in fun and fellowship – while giving back to the community.

Keegan Manz is a young boy living in nearby Morrisville, North Carolina, who was diagnosed with cystinosis in 2012 when he was six months old. When Tom met Keegan he was immediately drawn to his bright smile and laugh, his energy and life. During that first meeting with Keegan and his mom, Nicole, Tom recalled the hardships his father underwent with a similar genetic disorder, Alpha-1 antitrypsin deficiency.

Although Tom has no direct relationship with anyone with cystinosis, he was able to empathize with caring for a family member with a rare disease: the lack of resources to combat the disease; the education needed to understand the condition; the doctors' appointments; the round-the- clock medication schedule; the nights of worry; and worst of all, the uncertainty of survival. Tom remarked how Keegan's spirit and courage had shown through, without letting cystinosis get him down and without ever giving up.



Inspired by young Keegan, Tom began a series of events titled *A Cure for Keegan,* which has grown from three events in the Raleigh area to other statewide partnerships.

Keegan's cause has now been taken up by the Help A Brother Out Foundation (H.A.B.O.), where Tom serves as vice president.

He has also fostered partnerships with a Charlotte-area poker league, *Knockout Poker,* which is "adopting" a local cystinosis child and championing the family's fight against this disease.

Tom not only provides information about cystinosis to these organizations, but his expertise helps to ensure that they become effective fundraisers. To date, three more cystinosis events have been planned this year.

Felipe Wilkie, president of H.A.B.O. (left), and Brad Manz holding his son Keegan with other H.A.B.O. members

Tom saw a void in championing lesser-known causes, especially at the community level. He believes there is a genuine desire to help individuals and families in local neighborhoods and towns, even when community members don't know the family personally or don't have the time to formally volunteer.

"I believe everyone deserves a little help," says Tom. "If we can build our communities around knowing and helping our friends and neighbors, then no one has to shoulder their problems alone. We can all carry a piece of the burden and make life better for everyone."

If you have interest in supporting Tom, *Knock-out Poker* or the H.A.B.O. Foundation – or know of other organizations that would like to partner with them in their fight against cystinosis – contact him at knockoutpoker.RDU@gmail.com.

KNOCKOUT POKER USA

How the Blended Diet Changed Keegan's Life

By Nicole Manz, Keegan's mom, Morrisville, NC

Keegan is two years old and the blended diet has been the biggest game-changer in his life with cystinosis!

When Keegan was diagnosed with cystinosis, one of the hardest things for me, was when he stopped eating and drinking. The thought of a G-tube was scary. We tried for five months to get him to eat and take his medications by mouth, but he just couldn't do it. We hooked him up to a pump to provide the calories and fluids he needed, but he would vomit four or five times a day. We spent most of our time trying to prevent the vomiting.

A year ago when Keegan was one, we decided to start blending real food and putting it through his G-tube. We faced resistance from our medical team but the decision was not negotiable. Keegan needed real food in his tummy. Food would help keep the vitamins and medicines down.

My goal was to provide Keegan with a healthy diet of nuts, beans, grains, fruits and vegetables. Within several weeks we transitioned him completely onto the blended diet and water – and we noticed some amazing changes. Keegan's appetite increased; his nails began to grow faster; his height and weight increased; and he started to walk and make more sounds. But the biggest change – **he stopped vomiting.**

It has been almost a year since we started the blended diet, and Keegan has rarely vomited, except when he was sick with a cold. We have been able to take him off his reflux medication and Zofran, which prevents vomiting.

It was difficult to get Keegan down to three meals of blended food a day through his G-tube, but we are glad we did. It trained his stomach to follow "normal" eating patterns: tummy full … tummy empty … hungry … tummy full … tummy empty … hungry.

With the blended diet his body has learned how to digest real food versus formula. As children begin to eat on their own after months or years of tube feedings they get upset stomachs, which can cause them to associate eating with discomfort. Their bodies have to learn how to process real food.

Since the blended diet is given through the G-tube, you can transition at a rate that works best for your child. The blended diet sets children up to eat on their own! After a year on the blended diet, Keegan is eating completely on his own.

I love getting emails from parents in our community telling me how many improvements they've seen in their child's health since starting this diet! It may be something you would like to consider for your child.

How do I get started?

Every child is different and there are many ways to introduce real foods to your child through their G-tube. It can be as simple as buying packets of baby food or blending meals on your own. We have created a Facebook support group for parents who want to learn more about this diet. You can ask questions, get advice and find out how others have started. To learn more, search "real food for tubies with cystinosis."

How can I add calories to my blends?

There are several ways to add calories without making your blend too thick. You can add coconut oil, molasses, chia seeds, nuts, olive oil and tahini. Use nuts and seeds only if using a high-power blender such as a Vitamix or Blendtec.

What is a good blend when my child is sick or having a hard time keeping down medications?

My "go to blend" for an upset tummy is:

- 6 ounces of milk (formula, whole milk, goats milk or hemp milk all work fine. (Milk can upset the stomach, so it may help to just use water.)
- 2 bananas
- 1/2 cup of rolled oats
- 1 tablespoon of coconut oil
- 1 tablespoon of apple cider vinegar

Just blend and keep in the refrigerator. Use all day, just a few minutes before giving any medicine.

A Journey To Where Dreams Really Do Come True

By Jody Strauss, Gabbie's mom, Waterloo, Ontario, Canada

Gabbie (center) with friends (l–r) Kate Preis, Avaleigh Hansen, Kiley Moore, Leah Morrison

On March 27, more than 300 people attended *The Wizard of Oz* at the Dunfield Theatre in Cambridge, Ontario in support of Gabbie's Wish to cure cystinosis. The event raised $51,000! Guests enjoyed a candy bar, photo booth, silent auction, meet and greet with the cast and hors d'oeuvres courtesy of Milestones Restaurant.

The event was organized by the talented team at Isabel Avery & Co and many wonderful volunteers. Special thanks to Gabbie's Grandma, Dianne Strauss and Gloria Deutschlander for their tireless effort and for recruiting sponsors.

Several businesses stepped up to become premier sponsors for the event. They deserve recognition for being true friends to Gabbie. Special thanks to Ball Construction, Blue Top Properties and Strassburger Windows and Doors. We had more than 25 community sponsors for the event. We are grateful to each business that opened their hearts to Gabbie's story.

In December, we welcomed a healthy new baby girl, Hannah, to our family. There was excitement and joy in our home, but also extra work and less sleep! We are so grateful for our network of friends and family who rolled up their sleeves and got to work to get the job done. We have amazing people in our life!

We have been fundraising for six years. We couldn't keep going without the continued support from our family, friends and community. Thank you to everyone who has volunteered, attended events and given generously to help find a cure for cystinosis. We believe our prayers will be answered.

Friends from Milestones Restaurant (l–r) Dan Smith, Jeremy Wall, Brian Lonergan, Andrew Figueiredo

Event organizers from Isabel Avery & Co with the Wicked Witch (l–r) Cailey McQuay, Carrie Lepage, Witch Carrie Heath, Krista Hunter

Hard at work (l–r) Gloria Deutschlander and Dianne Strauss (Gabbie's Grandma)

Thank you for journeying with us somewhere over the rainbow where our dream of finding a cure for cystinosis will come true.

Gabbie holding her new sister, Hannah

Photos courtesy of Kaptured Studios

Chance For Chase

Benefiting Cystinosis Research Foundation

By Danielle Chodakowsky, Chase's mom, Fort Myers, Florida

Chase was born on February 16, 2012. He weighed in at a whopping 8 pounds 12 ounces, and he was 21½ inches long. He was perfect, *or so we thought*.

Chase with his dad, Jason; mom, Danielle; and sister, Taylor

The first year of Chase's life was uneventful. He was in the 75 percentile for his height and weight until he was about 15 months old. At that point, we had been to the pediatrician several times because we were concerned about his excessive thirst. We were told on multiple occasions that we were worrying too much and that he was just a "thirsty boy!" We even took him to a second doctor and were told the same thing.

In September 2015, we demanded that more tests be run because now, in addition to his excessive thirst, Chase began to refuse food. He was thin and frail, and we knew something was wrong. After being sent to multiple specialists and staying in the hospital for several nights while they ran tests, Chase's nephrologist diagnosed Chase with Fanconi syndrome and waited for blood work to confirm a diagnosis of cystinosis.

When the diagnosis was made, our world crumbled! Within days of Chase's diagnosis, we were contacted by Nancy Stack, president of the Cystinosis Research Foundation (CRF). We spoke for hours and she answered every question we had and eased all of our concerns. She gave us such an incredible feeling of hope, we felt that everything was going to be OK.

We knew that CRF was dedicated to funding research needed to find better treatments and ultimately a cure for Chase and all the other children and adults with cystinosis.

We wanted to help immediately but it took us about a year to get adjusted to our new life with cystinosis before we began planning our first fundraising event.

On February 7, 2015 we held our First Annual *Chance for Chase* Benefit. We sold tickets to the event, which included a delicious dinner prepared by the chef at Pelican Preserve; an amazing silent auction and raffle with more than 70 items up for grabs; entertainment by our live DJ; and gourmet cupcakes prepared by Custom Confections by Carry. Chase's nephrologist, Dr. Irina Gershin-Stevens, also spoke about cystinosis and its many complications.

About 160 guests attended that evening and raised an amazing $27,000 for cystinosis research. We are blessed to have such a supportive community. And we owe a million thanks!

It feels so good to be part of such an amazing foundation that will do everything possible to improve Chase's life, along with the many others who have cystinosis!

Thank you CRF for always being there for us and never allowing us to feel alone on this journey!

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Many Hands Make Lite Work

TOGETHER WE ARE MAKING A DIFFERENCE

The third annual Music for Mary benefit concert took place on April 11, at the spectacular LeMay Classic Car Museum in Tacoma, Washington. The concert, held in honor of 6½-year-old Mary Head, featured a command performance by longtime Music for Mary supporter Chris Anderson.

When they weren't being dazzled by the talented young singer, guests had an opportunity to tour the private LeMay Family Car Collection and participate in spirited silent and live auctions.

Special guests included eight-year-old twins, Emma and Gracie Patterson, and their wonderful grandma, Midge Patterson. Emma and Gracie, from nearby Federal Way, Washington, also have cystinosis. Grandma Patterson created a handmade quilt for the event. She sold raffle tickets for several months prior to the event and at the door.

The raffle for the beautiful quilt netted $1,785, bringing the evening's total to $14,315 for cystinosis research.

There was also a special check presentation from Head family friend, Karla Nore, who raised $485 at her work.

Thank you to everyone who worked on the event and to everyone who attended, making it another night to remember for cystinosis research.

Another Huge Success for Tina's Hope for a Cure

The 7th Annual Wine, Stein & Dine was held on Saturday, May 16, 2015. This annual celebration took place in the Rogers Scion Toyota showroom in Lewiston, Idaho, for the third year in a row. And once again it was a spectacular success.

More than 235 guests attended the special evening that included international cuisine and entertainment by Duet Riendeau. More than $110,000 was raised during the incredible silent auction and Fund a Cure. Thank you to everyone who volunteered, donated or attended.

Every dollar raised for Tina's Hope for a Cure Foundation goes directly to the Cystinosis Research Foundation to fund research that will lead to better treatments and a cure.

Congratulations to Denice and Mark Flerchinger and their family and friends who worked tirelessly to ensure that the event was an outstanding success!

www.tinashopeforacure.org.

Hearts for Hadley

Raising awareness and money for cystinosis has become a family affair! On March 7, Ben and I traveled with Hadley, and her big sister Stella, to the small town of Cove, Oregon to attend a volleyball tournament benefiting Hearts for Hadley.

Ben's cousin, Taylor Little, organized the tournament with help from her mother. Taylor approached us several months ago with the idea of doing something to help raise money for cystinosis. A high school senior, Taylor decided to combine her love for both Hadley and volleyball and coordinated this event as part of her senior project.

The all-female tournament took place at the Cove High School gym. Holly, Taylor's mom, was on-hand to sell snacks, Hearts for Hadley t-shirts and raffle tickets for gift baskets. It was amazing to see such a great turn out for the tiny town of only 550 people.

Taylor and her efforts raised more than $1,000 for Hearts for Hadley and CRF. Taylor also developed a presentation for her senior class explaining cystinosis and sharing a little bit about Hadley's daily life.

WE'VE GOT YOU COVERED

Our family moved to Boise, Idaho during the summer of 2013. Since then, we have been welcomed with open arms by our new hometown. Our daughters, Stella and Hadley, are fortunate they get to grow up in a place that embraces community and one that makes an effort to support local causes.

David Andrews, an incredible local musician, was featured as the first artist in a new music series that kicked off at the end of February this year. David chose Hearts for Hadley as the charity to benefit from the event. This is the second time the David Andrews Band has donated their time, energy and talents to raise money for Hearts for Hadley. David and his band performed a private concert last fall that raised $5,000, which was donated to CRF.

We've Got You Covered is a music series performed and recorded at Visual Arts Collective in Boise six times a year. Each show features a local musical artist and benefits a charity of their choice. The featured artist picks other musicians to cover one of their songs and the night ends with the featured artist performing a set.

The first concert was held on February 28, which was also Rare Disease Day. We attended the sold-out show along with many friends and family. The evening was incredible and helped raise over $2,000 for cystinosis research!

A version of this story originally appeared at www.erietvnews.com on March 27, 2015

Spa Fundraiser for a Boy with a Rare Disease

For the first 18 months of Jake Krahe's life, he was just like every other child. When he went to the hospital with some unusual symptoms in July of 2008, everything in Jake and his family's life changed. Jake, who lives in Medina, Ohio, with his parents and his twin brother, Austin, was diagnosed with a rare disease called cystinosis.

Cystinosis affects about 500 people in the U.S. and fewer than 2,000 worldwide. There are treatments for cystinosis, but there is no cure. As with any disease there are challenges, says Amy Krahe, Jake's mother. "Staying on his 24-hour-a-day medication schedule is critical and tough. We must get up in the middle of the night to give Jake his medication, and there are eye drops that need to be given every hour when he's awake."

Thankfully, a longtime friend has stepped up to help. Karen Staab wanted to find a way to celebrate her birthday, while helping Jake. Karen's goal was to raise awareness for the disease. "There are a lot of well-known causes but because cystinosis is so rare, there isn't much support for it or a lot of money for research," says Karen.

Amy Krahe with Karen Staab

But in her community there certainly was support. More than 50 people turned out for an evening of pampering at the Nature's Beauty Solutions salon in Erie, PA. Guests had their choice of a manicure, pedicure, facial or even an artistic henna tattoo.

Four students from the Academy of Cosmetology generously volunteered to provide services for the event. And Nature's Beauty Solutions also played an important role in this exciting day. The salon gave 60 percent of all the money raised to Jake's foundation.

When Jake's grandmother was asked what this all meant to her, she said, "It's overwhelming. Something like this for Jake, for my son, for Amy – I can't thank everyone enough."

Through it all, Jake, doesn't let cystinosis keep him down. Karen says, "He's a typical little guy, full of life." His mother adds, "If you ask anyone who knows Jake, they'll tell you that he always smiles from ear to ear." And when she thinks about Jake's future, she says, "We try to live for today and not worry about what tomorrow might bring."

If you'd like to help Jake, visit www.cystinosisresearch.org/donate-for-jake/

LITTLE TOWN. BIG HEART.

Saugeen Shores is a picturesque village in Bruce County, Ontario, Canada with a permanent population of about 7,500. Located on the Lake Huron coastline, the historic town is well known for its great beaches, campgrounds, lake cottages and spectacular sunsets. But today, perhaps Saugeen Shores is best known, at least in some circles, for its most famous resident, beautiful and ever-smiling Olivia Little.

Since they first heard about five-year-old Olivia and her diagnosis with cystinosis, the citizens of Saugeen Shores have grown to love her and want to help her, as well as all the others who have the disease. The Saugeen Shores community keeps coming up with innovative, and highly successful, ways to raise money for the Liv-A-Little Foundation and cystinosis research.

The temperature was below zero on the cold but sunny Saturday, April 4, morning but that didn't stop families from flooding the Saugeen Golf Club for the 4th Annual Liv-A-Little Egg-Stravaganza. The fun-filled event featured games such as bunny hop, egg rolls, pin the tail on the bunny and a special visit from the Easter Bunny himself. The hunt was sold out with over 250 people attending and raising more than $2,000 for cystinosis research.

LIV-A-LITTLE FOUNDATION

Just five weeks later there was another creative event for Olivia.

What little girl doesn't want to be a Princess for a Day?

On May 9, Sinless Lash, in downtown Saugeen Shores, helped that dream come true for many little girls. Olivia and her royal entourage filled the salon as they got their nails painted, and their hair curled and braided. Of course, they also received sparkling tiaras and magic wands as the all-important finishing touches. The event was a regal success raising more than $500 for cystinosis research.

Who could ask for more? *An event that truly helped make two dreams come true: a Princess for a Day and a cure for cystinosis.*

The little town of Saugeen Shores, Ontario has a very big heart, and obviously loves Olivia, its little princess, and all her friends from around the world who have cystinosis.

Erin and Chad Little, are enormously thankful for the generosity they continue to receive from their family and friends in the Saugeen Shores community. The support of this small town with its big heart means that Olivia can truly look forward to a life free of cystinosis.

Ruth Ann Ahnen (r), reached the summit of Africa's highest peak, Uhuru Peak on Mount Kilimanjaro, on January 8. She is shown with climbing partners Jane Doyon (l) Kathy Warren (center)

Climbing to New Heights for Cystinosis

A version of this story originally appeared in the online edition of the Milwaukee Journal Sentinel, *February 12, 2015*

On January 8, 2015, local swim coach Ruth Ann Ahnen summited Mount Kilimanjaro to raise money for the Cystinosis Research Foundation. Kilimanjaro's Uhuru Peak, which Ahnen climbed, is Africa's highest point, at nearly 20,000 feet.

Ruth Ann coaches the Arrowhead High School's boys and girls swim teams in Hartland, Wisconsin and the Lake Country School swim team. Her daughter Katie, 24, lives with cystinosis, a rare metabolic disease that affects about 500 people in United States and fewer than 2,000 worldwide.

Ahnen has climbed to Mount Everest Base Camp and run the New York Marathon to raise money to fight cystinosis in the past. Her latest feat was nearly derailed by weather and altitude issues.

"We could not have summited without our guides or each other, and without the constant reminder of who I was climbing for," Ahnen said in a news release.

One hundred percent of the proceeds from the climb will go to the Cystinosis Research Foundation.

Wear Your Rare

Wear Your Rare began as a simple dare between a father and son, Clinton and Chandler Moore, who has cystinosis, to see who could stand outside in their underwear the longest on a freezing cold night. After posting their picture on Facebook, several people suggested that they should do it as a fundraiser.

Three cystinosis parents – Jennifer Bartkowski, Nicole Manz, and Clinton Moore – adopted the idea and created the Wear Your Rare challenge for their families, friends and neighbors.

The plan ... to wear something extremely rare – the funnier the better – and go outside for 17 minutes. During those 17 minutes you need to snap a picture and post it to the Wear Your Rare Facebook page. Then donate $17 to the Cystinosis Research Foundation to help fund research for cystinosis. The final step: challenge 17 friends to do the same.

Why 17 of everything? Cystinosis is linked to a mutation or deletion of chromosome 17; and many cystinosis patients must take at least 17 different medications and pills everyday to maintain their health. Cystinosis patients must also use eye drops every hour they are awake, which often equals 17 hours a day!

As of publication, Wear Your Rare has raised $1,913! Thank you for helping us find better treatments and ultimately find a cure for cystinosis. **To donate or participate, visit the Wear Your Rare Facebook page.**

Kuepfer Family Snaps the Lid for Cystinosis

This spring, five-year-old Amanda Kuepfer and her mother, Rachel, from Millverton, Ontario, Canada, held a Tupperware fundraiser. With support from family and friends, they were able to raise $1,096, which the Kuepfers sent to CRF in Amanda's honor.

Be sure to watch the fall issue of *Cystinosis Magazine* for news about the Amanda's Hope for a Cure event that was held on May 30.

it takes a village
WITH LOTS OF ACTIVITIES

Thursday, July 2, 2015
Dinner with Ben Stein
24 Hours for Hank,
Henry Sturgis
Trinity at City Beach, Idaho
Tickets: **tricia.sturgis@gmail.com**

Sunday, August 23, 2015
Ironman Triathlon by Trevor Theobald,
in honor of Jake Krahe
Grand Rapids, Michigan
To contribute:
www.cystinosisresearch.org/donate-for-jake

Saturday, September 12, 2015
4th Annual
Fore Fathers JCFG
Golf Tournament
Andrew Cunningham
Boulder Creek Golf Course, Langdon, Alberta, Canada
Information, contact Karen McCullagh at:
kcm_consulting@msn.com

Sunday, September 20, 2015
Morgan Peachman
Bob-O-Link Golf Course
Avon, Ohio
www.cystinosisresearch.org/mulligans-fore-morgan

Friday, September 25, 2015
Hearts for Hadley,
Hadley Alexander
Information:
Hearts for Hadley Facebook page

Saturday, November 7, 2015
Lots of Love for Landon Halloween Event,
Landon Hartz
Castle Shannon Fire Hall, Pittsburgh, PA
Information: **laurenhartz@gmail.com**

Saturday, November 7, 2015
Dirt Bike Ride for a Cure,
Bailey DeDio
Barstow/Stoddard Valley
(Off-Highway Vehicle Area)
Information or tickets:
www.cystinosisresearch.org/donate-for-bailey

Dirt Bike Ride for a Cure

March 10 and March 11, 2016
Swing & Bling Fundraiser,
Jenna and Patrick Partington
Thursday, Golf: Catta Verdera Golf Club
Friday, Dinner Gala: The Citizen Hotel
Information: **www.jennaandpatrick.org**

March 2016
24 Hours of Schweitzer Ski Event
24 Hours for Hank,
Henry Sturgis
Schweitzer Mountain, Sand Point, Idaho
Information: **www.24hoursforhank.org/events.html**

April 7 – April 9, 2016
CRF DAY OF HOPE Family Conference
New location: Island Hotel
Newport Beach, California

Information: Nancy Stack
nstack@cystinosisresearch.org

cystaran™
(cysteamine ophthalmic solution) 0.44%

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1

***To order**, please call:*
Walgreens Specialty Pharmacy
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2

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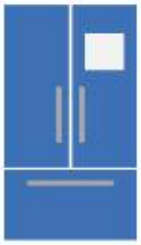

Store unopened bottles in the freezer

4

Every 7 days remove 1 bottle and thaw for 24 hours before use*

***NO REFRIGERATION NECESSARY AND DO NOT REFREEZE**

5

Discard bottle after 7 days

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*You are encouraged to report negative side effects of prescription drugs to the FDA. Visit **www.fda.gov/medwatch or call 1-800-FDA-1088***

SCIENTIFIC REVIEW BOARD

The Scientific Review Board 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 submitted and advising the CRF on the scientific merit of each proposal.

SCIENTIFIC REVIEW BOARD

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2015 CALL FOR RESEARCH PROPOSALS

In 2003, Nancy and Jeff Stack established the non-profit Cystinosis Research Foundation (CRF), with the goal of funding cystinosis research to find better treatments and a cure for cystinosis. Since its inception, CRF has raised more than $29.3 million, with every dollar raised going directly to cystinosis research.

CRF will announce its fall 2015 call for research proposals and fellowships in September 2015. Details and guidelines for applications will be available at: www.cystinosisresearch.org/research/for-researchers.

The Cystinosis Research Foundation utilizes a Scientific Review Board (SRB) comprised of leading international experts in the field of cystinosis. The SRB provides independent, objective reviews and recommendations for each research proposal submitted based on the NIH scale of standards. The SRB follows grant review guidelines established by the Cystinosis Research Foundation (CRF) and advises the CRF on the scientific merits of each proposal.

The goal of the Cystinosis Research Foundation 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 2010, CRF established the **Cure Cystinosis International Registry (CCIR)** to serve as a hub of information about cystinosis and its complications. Currently CCIR has approximately 460 registrants in 40 countries around the world. The site contains a Professional Research Portal for researchers who register to access and view de-identified, aggregate cystinosis patient information. **(www.cystinosisregistry.org)**

CRF is excited about the future of cystinosis research, and is grateful to its researchers for their interest in the cystinosis community and its children. 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 56 studies, including these studies, which were published since the fall 2014 *Cystinosis Magazine*.

- Lysosomal Targeting of Cystinosin Requies AP-3 – Published online March 2015 in *Traffic* by Corinne Antignac, MD, PhD, Professor, Imagine Institute, Laboratory of Hereditary Kidney Diseases, Paris, France.
- The Swan-Neck Lesion: Proximal Tubular Adaptation to Oxidative Stress in Nephropathic Cystinosis – Published online February 18, 2015 for the *American Journal of Physiology* by Robert L. Chevalier, MD, Department of Pediatrics, University of Virginia.
- Ocular Drug Delivery Nanowafer with Enhanced Therapeutic Efficacy – Published online February 6, 2015 at ACSNANO by Ghanashyam Acharya, PhD, Baylor College of Medicine.
- Impairment of Chaperone-Mediated Autophagy Leads to Selective Lysosomal Degradation Defects in the Lysososmal Storage Disease Cystinosis – Published January 2015 in *EMBO Molecular Medicine* by Sergio D. Catz, PhD, The Scripps Research Institute.
- Clinical Utility of Chitotriosidase Enzyme Activity in Nephropathic Cystinosis – Published September 30, 2014 in *Orphanet Journal of Rare Diseases* by Elena Levtchenko, PhD, University of Leuven, Belgium.

2014 CRF Research Grants Funded • Total: $2,068,247

Ghanashyam Acharya, PhD, Principal Investigator
Baylor College of Medicine, Houston, Texas
Jennifer Simpson, MD, Co-Principal Investigator
University of California, Irvine
"Development of Extended Release Cysteamine Nanowafer"
$276,380 – 2-year grant
(September 1, 2014 – August 31, 2016)

Corinne Antignac, MD, PhD, Principal Investigator
Inserm U1163
Imagine Institute, Paris, France
"Characterization of the mTORC1 Pathway in Cystinosis"
$266,000 – 2 year grant
(February 1, 2015 – January 31, 2017)

Anand Bachhawat, PhD, Principal Investigator
Indian Institute of Science Education and Research (IISER), Nagar, Punjab, India
"Genetic and Biochemical Studies on the Cystinosin Transporter Using a Novel Genetic Screen"
$82,500 – 2-year grant
(September 1, 2014 – August 31, 2016)

Sergio Catz, PhD, Mentor
Jinzhong Zhang, PhD, Fellow
The Scripps Research Institute, La Jolla, California
"Improvement of Cellular Function Through Chaperone-Mediated Autophagy and Cellular Trafficking in Cystinosis"
$150,000 – 2-year grant
(September 15, 2014 – September 14, 2016)

Pierre J. Courtoy, MD, PhD, Principal Investigator
Héloïse Chevronnay, PhD, Co-Principal Investigator
Christophe Pierreux, PhD, Co-Principal Investigator
de Duve Institute, UCL, Brussels, Belgium
"Testing Endocytosis-Based Injury with Double Megalin/Cystinosin KO Mice and Exploring Stem Cell: Epithelial Connectivity Across Basement Lamina"
$246,272 – 2-year grant
(September 1, 2014 – August 31, 2016)

Alan Davidson, PhD, Principal Investigator
Teresa Holm, PhD, Co-Principal Investigator
The University of Auckland, Grafton, Auckland, New Zealand
"Cystinotic iPCSs: Generation of Proximal Tubule Cells and Role of the Malate-Aspartate Shuttle"
$207,757 – 2-year grant
(September 1, 2014 – August 31, 2016)

Olivier Devuyst, MD, PhD, Mentor
Alessandro Luciani, PhD, Fellow
Institute of Physiology, UZH, Zürich, Switzerland
"Lysosomal Function, Autophagic Clearance and Junctional Integrity in Nephropathic Cystinosis"
$225,000 – 3-year grant
(September 1, 2014 – August 31, 2017)

Francesco Emma, MD, Principal Investigator
Bambino Gesù Children's Hospital, IRCCS, Rome, Italy
"In Vivo and In Vitro Drug Screening and Testing for Nephropathic Cystinosis"
$214,665 – 2-year grant
(September 1, 2014 – August 31, 2016)

Bruno Gasnier, PhD, Mentor
Xavier Leray, PhD Candidate, Fellow
Université Paris Descartes, Neurophotonics Laboratory/CNRS, Paris, France
"Mechanism and Modulation of Cysteamine Therapy"
$150,000 – 2-year grant
(October 8, 2014 – October 7, 2016)

Mary Leonard, MD, Principal Investigator
Paul Grimm, MD, Co-Principal Investigator
Stanford University School of Medicine, Stanford, California
"Musculoskeletal Disease in Children and Adults with Cystinosis"
$174,339 – 1-year grant
(August 1, 2015 – July 31, 2016)

Aude Servais, MD, PhD, Principal Investigator
Nathalie Boddaert, MD, PhD, Co-Principal Investigator
Necker Hospital, Paris, France
"Neurological Complications in Cystinosis Patients"
$50,034 – 1-year grant
(June 1, 2015 – May 31, 2016)

Doris Trauner, MD, Principal Investigator
University of California, San Diego
"Academic Achievement and Quality of Life in Individuals with Cystinosis"
$25,300 – 2-month extension grant
(December 1, 2014 – January 31, 2015)

Ghanashyam Acharya, PhD, Principal Investigator
Baylor College of Medicine, Houston, Texas
Jennifer Simpson, MD, Co-Principal Investigator
University of California, Irvine

"Development of Extended Release Cysteamine Nanowafer"
$276,380 – 2-year grant (September 1, 2014 – August 31, 2016)

Objective/Rationale: Corneal cystinosis is presently treated with cysteamine eye drops. Hourly administration of the eye drops several times in a day is often required to treat corneal cystinosis, which is very difficult to comply with for infants, school going children, and young adults. Due to multiple administrations on a daily basis, patients develop side effects such as excessive tearing, redness, and ocular inflammation. Consequently, compliance and treatment outcome are severely compromised. This research project focuses on the development of an extended release cysteamine nanowafer to treat corneal cystinosis.

Project Description: This research effort aims to develop an extended release cysteamine nanowafer that can release cysteamine for up to a week with negligible side effects. The nanowafer is a tiny disc (5-8 mm diameter) that can be applied on the cornea with a fingertip. The nanowafer will slowly dissolve and fade away at the end of the stipulated drug release time. The nanowafers will be fabricated by electron-beam lithography and the drug will be loaded by microinjection. The efficacy of the cysteamine nanowafer will be evaluated in cystinosine knockout mice by quantifying the total cysteine concentration in the nanowafer treated group and compared with that of an untreated group. A successful outcome of the preclinical studies will provide a strong evidence-based rationale for the translation of nanowafer drug delivery systems to clinical trials in humans for treating corneal cystinosis.

Relevance to the Understanding and/or Treatment of Cystinosis: The presently available treatment for corneal cystinosis is cysteamine eye drops. The eye drops although effective, require multiple dosings per day and cause serious side effects. The extended release cysteamine-nanowafer can surmount these issues and improve patient compliance with the treatment. Upon successful development, the cysteamine-nanowafer will be a major advancement in the corneal cystinosis treatment.

Anticipated Outcome: Upon successful completion of the project, a clinically translatable csyteamine-nanowafer drug delivery system will be developed.

Francesco Emma, MD, Principal Investigator
Bambino Gesù Children's Hospital, IRCCS, Rome, Italy

"In Vivo and In Vitro Drug Screening and Testing for Nephropathic Cystinosis"
$214,665 – 2-year grant (September 1, 2014 – August 31, 2016)

Objective/Rationale: Despite very significant improvements over the past decades, treatment for cystinosis remains sub-optimal and new therapies are needed. To this end, we have begun screening a drug library for compounds that can decrease cystine content or the rate of cell death (apoptosis) in cystinosis cells.

Project Description: In this project, we will test directly in cystinotic mice the benefits of one drug that was identified in our previous screening. This drug will be tested in alternative or in combination with cysteamine. In addition, we will use newly identified characteristics of cystinotic cells to search for additional drugs that may be beneficial to patients.

Relevance to the Understanding and/or Treatment of Cystinosis: This application is aimed at improving treatment of nephropathic cystinosis by testing directly in animal models one drug that has the potential of being useful in treating this disease and at searching additional candidate molecules.

Anticipated Outcome: Should the results of these tests be positive, they may lead to directly testing new therapies in human subjects.

Corinne Antignac, MD, PhD, Principal Investigator

Inserm U1163
Imagine Institute, Paris, France

"Characterization of the mTORC1 Pathway in Cystinosis"
$266,000 – 2-year grant (February 1, 2015 – January 31, 2017)

Objective/Rationale: As cysteamine does not correct all symptoms of cystinosis (i.e. Fanconi syndrome), we hypothesized that cystinosin, the protein altered in cystinosis, could have a novel role, in addition to transporting cystine out of the lysosome. Mass spectrometry enabled us to identify cystinosin as a component of the mTORC1 pathway, a system that controls organ growth and metabolism. We further showed that the mTORC1 pathway activity is decreased in renal proximal tubular cells derived from Ctns-/- mice and that these alterations are due to the absence of cystinosin and not to cystine accumulation, as the decrease of lysosomal cystine levels by cysteamine does not rescue mTORC1 activation. Our results clearly show a dual role for cystinosin both as a cystine transporter and as a component of the mTORC1 pathway.

Project Description: Our project for the next two years is to extend our previous studies from a cell model to the Ctns-/- cystinosis mouse model and to better dissect the role of cystinosin in the mTORC1 pathway. We thus will (i) characterize the mTORC1 pathway in the Ctns-/- mouse model; (ii) further characterize the role of cystinosin in the amino acid sensing machinery by assessing its role in the interactions between the various members of the mTORC1 pathway; (iii) use the new and very promising Crispr/Cas9 technology to create cell lines with CTNS mutations and check in these cell models the activity of the mTORC1 pathway.

Relevance to the Understanding and/or Treatment of Cystinosis: If our results are confirmed, they will definitely prove that cystinosin function is more complex than just transporting cystine out of the lysosome and that it is necessary to develop new treatments not based only on cystine depletion.

Anticipated Outcome: We expect to show that the mTORC1 pathway is altered in the cystinosis mouse model.

We also want to create new tools (especially, a cell line with endogenous "tagged" cystinosin to be able to follow easily its behavior in cells, which is not possible until now, given the lack of good antibodies against cystinosin) that will be helpful for the whole cystinosis research community.

Anand Bachhawat, PhD, Principal Investigator

Indian Institute of Science Education and Research (IISER), Nagar, Punjab, India

"Genetic and Biochemical Studies on the Cystinosin Transporter Using a Novel Genetic Screen"
$82,500 – 2-year grant (September 1, 2014 – August 31, 2016)

Objective/Rationale: The cystinosin protein (CTNS) is a lysosomal membrane transporter that transports cystine. The current methods used to evaluate the function of CTNS are cumbersome. This has impeded investigations directed toward finding out the functionally critical parts of the protein. We would like to develop a simpler method to functionally evaluate the protein so that we can subject it to detailed genetic/mutational analysis. The new assay would be yeast-based and would allow one to rapidly evaluate and investigate cystinosin function.

Project Description: Efforts would be made to evaluate yeast as a host to functionally evaluate the human cystinosin transporter. CTNS would be expressed in yeast and the function evaluated by a simple and inexpensive plate assay that would allow one to isolate mutants of the protein for functional evaluation. Mutagenizing a protein followed by functional evaluation is the key to understanding protein function. The yeast method would allow one to do such a mutagenesis – both through a random strategy and a site-directed strategy- and quickly evaluate their function. To confirm the validity of the findings, the function of the protein and the mutants would be compared with their function when expressed in the mammalian system. Using this approach the substrate-binding channels and other functionally important domains could be delineated.

Relevance to the Understanding and/or Treatment of Cystinosis: As cystinosis is caused by a defect in CTNS, it is important to have an in-depth understanding of CTNS as it would enable one to understand how the different kinds of mutations (in CTNS) found in cystinosis patients would impact the functioning of the transporter, and the disease manifestation. Different levels of functioning might require different kinds of treatment. This in-depth understanding of CTNS is currently lacking. We hope that the method developed here would greatly accelerate our understanding of the protein and thereby have an impact on the treatment.

Anticipated Outcome: The method developed for functional evaluation of CTNS would facilitate and augment the efforts on describing the critical features of the protein in terms of substrate-binding channels and other functional domains. It is also possible that the method developed could be eventually used as a diagnostic assay in patients for cystinosin protein function.

Sergio Catz, PhD, Mentor
Jinzhong Zhang, PhD, Fellow

The Scripps Research Institute, La Jolla, California
"Improvement of Cellular Function Through Chaperone-Mediated Autophagy and Cellular Trafficking in Cystinosis"
$150,000 – 2-year grant (September 15, 2014 – September 14, 2016)

Objective/Rationale: Mammalian cells contain intracellular compartments intended to degrade macromolecules and then recycle some small components back to the main soluble compartments. These components are then utilized to synthesize new macromolecules. In this way, mammalian cells eliminate unwanted components while saving energy and resources by maintaining a constant supply of essential elements. Degradation takes place in vacuoles denominated lysosomes (Greek roots: luo means "to destroy" and soma means "body".)

In cystinosis, some essential degradative products cannot be recycled and remain in the lysosomes. This induces lysosomal malfunction, lack of resources, accumulation of degradative products, cell malfunction and cell death. We found that one of the specialized lysosomal functions, named chaperone, mediated autophagy (CMA) is defective in cystinosis. We propose to study the mechanisms of CMA and to develop strategies to improve cell function in cystinosis.

Project Description: We found that the expression of an important regulatory protein named LAMP2a is decreased in cystinosis. LAMP2a is the only known receptor for chaperone-mediated lysosomal degradation. Defective CMA leads to the accumulation of toxic substrates and is involved in the pathogenesis of human diseases including kidney pathologies, neurological disorders, cancer and aging.

We will utilize cystinotic cells from both mouse models and humans with cystinosis to a) study the interplay between CTNS protein and the CMA receptor LAMP2a, b) understand the mechanisms of defective LAMP2a downregulation and mislocalization in cystinotic cells, c) understand the mechanism of defective translocation of substrates for degradation into the lysosomal lumen and d) determine the molecular basis of the regulation of CMA activity by CTNS.

We recently showed that increasing the movement of lysosomes in a cell, facilitates its function by increasing the probability of interaction with regulatory components, in the same way that public transportation enhances the function of a city by facilitating access of citizens to different working areas increasing productivity. We will express trafficking proteins to correct LAMP2a distribution and function. Finally, we will check the hypothesis that the accumulation of degradative products in lysosomes affects LAMP2a function. We will decrease lysosomal overload and study the role of CTNS mutants on the localization and function of LAMP2a and in the process of chaperone-mediated degradation in cystinotic cells.

Relevance to the Understanding and/or Treatment of Cystinosis: Defective CMA is directly linked to human disease, including kidney pathologies, an organ in which CMA is markedly active. Our research is highly relevant because it identifies, in cystinosis, previously unrevealed cellular defects associated with human pathologies. Elucidating the mechanisms that lead to abnormal CMA in cystinosis and determining strategies to rescue this phenotype will lead to a better understanding of the physiopathology of this disease and to novel approaches for the treatment of cystinosis.

Anticipated Outcome: The aim of our study is to discover why cystinotic cells develop CMA defects and how this impairment can contribute to the pathogenesis of cystinosis. Importantly, we will use different approaches aimed at ameliorating these cellular defects and improving cell function. We expect that our approach will lead to a better understanding of the pathogenic events in cystinosis and to the development of new strategies to improve cell function, which is fundamental to define novel treatments for cystinosis.

Doris Trauner, MD, Principal Investigator

University of California, San Diego
"Academic Achievement and Quality of Life in Individuals with Cystinosis"
$25,300 – 2-month extension grant (December 1, 2014 – January 31, 2015)

The work to be completed during this time includes two specific aims: (1) To complete data analysis and manuscript completion for the adolescent quality of life study then submit the paper for publication and (2) To conduct data analysis on the PROMIS study, write the manuscript and submit it for journal publication.

Pierre J. Courtoy, MD, PhD, Principal Investigator • Héloïse Chevronnay, PhD, Co-Principal Investigator
Christophe Pierreux, PhD, Co-Principal Investigator
de Duve Institute, UCL, Brussels, Belgium

"Testing Endocytosis-Based Injury with Double Megalin/Cystinosin KO Mice and Exploring Stem Cell: Epithelial Connectivity Across Basement Lamina"
$246,272 – 2-year grant (September 1, 2014 – August 31, 2016)

Objective/Rationale: Our investigations aim at better understanding the natural course of cystinosis and mechanisms of correction by hematopoietic stem cells (HSCs) in cystinotic mice (cystinosin KO). We shall (i) test whether apical receptor-mediated endocytosis of ultrafiltrated plasma proteins is the key source of lysosomal cystine in kidney proximal tubular cells (PTCs), primarily injured in nephropathic cystinosis; (ii) define dynamics and molecular actors by which HSCs, attracted by injured PTCs, project as tunneling nanotube (TNTs)/invadopodia and cross tubular basement lamina barrier; and (iii) analyse structure of PTCs basement lamina in cystinosin KO and HSC-grafted cystinotic mice.

Project Description: (i) To test the role of apical receptor-mediated endocytosis, we are generating double KO mice that lack cystinosin in all tissues (cystinosin KO) and are engineered to be further defective for apical receptor-mediated endocytosis selectively in kidney by conditional inactivation of the cornerstone endocytic receptor, megalin (megalin KO). Kidney protection in double KO mice will be evaluated by reference to single cystinosin KO and single megalin KO mice. Lack of protection in other cystinotic organs will serve as control. (ii) Dynamics and molecular machineries involved in TNT/invadopodia formation will be studied by vital multiphoton and high-resolution multiplex immunofluorescence in cystinotic mice engrafted with fluorescent HSCs. We shall focus on HSC polarization toward injured epithelia, cytoskeleton remodeling to support oriented protrusion, molecular motors guiding exocytosis and proteases digesting basement lamina. (iii) Basement lamina structure will be defined by combination of fluorescence and electron microscopy.

Relevance to the Understanding and/or Treatment of Cystinosis: This project thus targets physiopathology of nephropathic cystinosis and mechanisms of stem cell therapy in the kidney.

Anticipated Outcome: These investigations should demonstrate a key role of endocytosis in initiation and progression of nephropathic cystinosis. We also hope to narrow down our understanding of cellular and molecular processes allowing epithelial correction by HSCs, so as to help optimize the benefit of stem cell therapy.

Alan Davidson, PhD, Principal Investigator • Teresa Holm, PhD, Co-Principal Investigator
The University of Auckland, Grafton, Auckland, New Zealand

"Cystinotic iPCSs: Generation of Proximal Tubule Cells and Role of the Malate-Aspartate Shuttle"
$207,757 – 2-year grant (September 1, 2014 – August 31, 2016)

Objective/Rationale: How cystine accumulation in the proximal tubule cells of the kidney causes renal damage in cystinosis is largely unclear. A major challenge to solving this problem is a lack of good laboratory models of cystinosis. To help overcome this, we have generated cystinotic stem cells (called induced pluripotent stem cells; iPSCs) that can form any cell type in the body. The objectives of this proposal are to convert cystinotic iPSCs into proximal tubule cells and use these cells to (1) identify differences between normal and diseased cells and (2) explore whether the malate-aspartate shuttle (a biochemical pathway involved in energy production) plays a role in cystinosis.

Project Description: Normal and cystinotic iPSCs will be matured into proximal tubule cells using a method we have developed. How well these cells resemble 'natural' proximal tubule cells will be determined by a 'molecular fingerprinting' approach and functional testing. This analysis will validate the usefulness of iPSC-derived kidney cells and also identify informative cystinosis-specific differences that may lead to new therapeutic targets. The levels of amino acids, such as aspartate and other metabolites, will be examined in cystinotic cells and the therapeutic potential of these factors explored.

Relevance to the Understanding and/or Treatment of Cystinosis: Existing laboratory models of cystinosis do not recapitulate all the defects seen in patients. Thus, there is a need for new sources of cystinotic cells to study. Our project will pioneer the use of iPSC-derived proximal tubule cells as a new tool to investigate the cause of kidney failure in cystinosis. Our analysis of amino acid levels and other metabolites may uncover new ways to treat cystinosis.

Anticipated Outcome: Differences between normal and cystinotic proximal tubule cells will be identified and supplementation of cystinotic cells with metabolites found to be depleted may help correct the cellular defects associated with cystinosis.

Olivier Devuyst, MD, PhD, Mentor • Alessandro Luciani, PhD, Fellow

Institute of Physiology, UZH, Zürich, Switzerland

"Lysosomal Function, Autophagic Clearance and Junctional Integrity in Nephropathic Cystinosis"

$225,000 – 3-year grant (September 1, 2014 – August 31, 2017)

Objective/Rationale: Nephropathic cystinosis is characterized by a generalized dysfunction of the proximal tubule that progresses, if untreated, to end-stage renal disease. By using a Ctns mouse model, we demonstrated that the loss of cystinosin function in proximal tubule cells triggers an abnormal transcription program with defects in the endolysosomal pathway leading to the urinary loss of specific ligands, before structural damage or renal failure. These abnormalities raise the question of the mechanism(s) linking lysosomal accumulation of cystine to tubular cell dysfunction associated with nephropathic cystinosis.

Project Description: The overall goal of this project is to take advantage of a detailed characterization of Ctns mouse in vivo, combined to cutting-edge cell biology approaches applied on primary cultures of proximal tubule cells to analyze the role of lysosomal dysfunction as a major pathogenic event in the early stage of cystinosis. The specific aims include: (i) to characterize the progressive "identity crisis" and dysfunction of the lysosomal network; (ii) to evaluate the lysosomal clearance of autophagic cargoes (e.g. autophagosome membrane LC3-II, ubiquitinated proteins and dysfunctional mitochondria); (iii) to investigate how the decline of autophagic-lysosomal clearance affects the integrity of the junctional complex proteins and leads to an abnormal transcriptional program causing proliferation and apical dedifferentiation of the proximal tubule cells.

Relevance to the Understanding and/or Treatment of Cystinosis: Obtaining insights into the role of lysosomal dysfunction in the early chain of events leading to proximal tubule cell dysfunction, before structural damage, should yield new therapeutic targets in order to reverse clinically relevant manifestations of nephropathic cystinosis.

Anticipated Outcome: These translational investigations will address the role of failure of lysosomal network in the early stage of disease, and may point to cellular pathways that could be targeted (or monitored) before any structural, irreversible damage of the kidney. The mechanisms identified in early cystinosis may also be relevant for other forms of tubular disorders, helping us to better understand the link between proximal tubule dysfunction and renal disease progression.

Bruno Gasnier, PhD, Mentor • Xavier Leray, PhD Candidate, Fellow

Université Paris Descartes, Neurophotonics Laboratory/CNRS, Paris, France

"Mechanism and Modulation of Cysteamine Therapy"

$150,000 – 2-year grant (October 8, 2014 – October 7, 2016)

Objective/Rationale: Cystinosis is caused by accumulation of the amino acid cystine in an intracellular organelle, the lysosome. Cysteamine, the main treatment of cystinosis, rescues this defect by converting cystine into a new compound that exits lysosomes through the newly discovered protein PQLC2. This role of PQLC2 in cysteamine therapy was established using a cellular model of cystinosis. We now aim to examine this process at the organismal level using a mouse model of the disease. Our project will also test whether the activity of PQLC2 can be modulated to potentiate cysteamine therapy.

Project Description: Drugs susceptible to modulate PQLC2 will be tested on purified lysosomes and in cellular models of cystinosis. The activity of PQLC2 will be measured using selective radiolabelled substrates. Alternatively, we will use an analytical technique (mass spectrometry) to follow the buildup and decay of the reaction product formed by cystine and cysteamine.

Cystinosis is caused by mutation of the CTNS gene. To assess the role of PQLC2in the whole animal, we will breed mice defective for the PQLC2 gene with the current mouse model of cystinosis, which is defective for the CTNS gene. This will generate mice carrying mutations in both genes. These mice will be characterized at biochemical, histological and behavioral levels and compared with the current model. In a second step, the two models will be treated with cysteamine and subjected to biochemical and histological analyses in various organs.

Relevance to the Understanding and/or Treatment of Cystinosis: The novel mouse model should tell us whether PQLC2 is involved in cysteamine therapy for all organs and could provide a molecular basis for differences in cystine depletion efficacy. The genetic inactivation of PQLC2 may also exacerbate the severity of cystinosis in the absence of treatment according to preliminary data obtained with cellular models. The search for PQLC2 modulators aims to improve cysteamine therapy.

Anticipated Outcome: As a result of our study, we expect to arrive at a molecular understanding for inter-organ and, possibly, inter-individual differences in the response to cysteamine therapy. Furthermore, the identification of PQLC2 modulators could open the way towards more effective pharmacological treatments of cystinosis.

Aude Servais, MD, PhD, Principal Investigator
Nathalie Boddaert, MD, PhD, Co-Principal Investigator
Necker Hospital, Paris, France
"Neurological Complications in Cystinosis Patients"
$50,034 – 1-year grant (June 1, 2015 – May 31, 2016)

Objective/Rationale: Although rare, central nervous system complications do occur in adults with cystinosis. By brain imaging, cerebral atrophy is observed in all patients with central nervous system symptoms, but also in patients without gross central nervous system clinical abnormality. By magnetic resonance imaging (MRI), children with cystinosis evidence selective changes in cerebral white matter in areas of the visual pathway. Our objective is to analyze neurological complications in late adolescent and adult patients with cystinosis in the era of early cysteamine treatment.

Project Description: Systematic neurological examination and multimodal brain MRI will be performed in a cohort of 20 cystinotic patients and compared to 20 controls.

Complete patient's clinical history will be recorded. Adhesion to cysteamine treatment will be graded. A complete clinical and neurological examination will be performed, including muscular testing and swallowing evaluation. A biological sample will be taken to assess renal function, Fanconi syndrome, thyroid function status, diabetes detection and cystine leukocyte level.

Computed tomography scan will be performed in patients to search and localize brain calcifications. High resolution three dimensional images by MRI will be used as well as specific sequences for evaluating tissue cellularity, white matter integrity, resting state activity, to search microhemorrragic lesions, and to see the vessels. Magnetic Resonance spectroscopy will search cystine peak and thus cystine accumulation.

Relevance to the Understanding and/or Treatment of Cystinosis: Little is known about the long-term progression of adult nephropathic cystinotic patients since cysteamine treatment became available, especially for neurological complications. Furthermore, the development of advanced neuroimaging techniques has provided new tools to investigate the underlying neurophysiopathological mechanisms of metabolic diseases. The characterization of the lesions will help the understanding of the pathogenesis of the neurological complications in cystinosis. We will be able to explore any vascular disease and to detect inflammatory lesions.

Anticipated Outcome: Our hypothesis is that there is a progressive development of brain abnormalities but early treatment with cysteamine would reduce the incidence of such complications. A progressive microvascular disease of the brain and a cerebral cystine crystal-associated vasculopathy might be detected. Systematic imaging would also show asymptomatic lesions. A better detection of these neurological complications could lead to early diagnosis, cysteamine treatment adjustment and more targeted interventions and follow up to reduce their negative impact in adulthood.

Cystinosis Research Foundation

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MISSION

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

On the eve of her 12th birthday,
Natalie Stack made a wish
no child should ever have to make

In 2003
CRF was Founded

100%
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60


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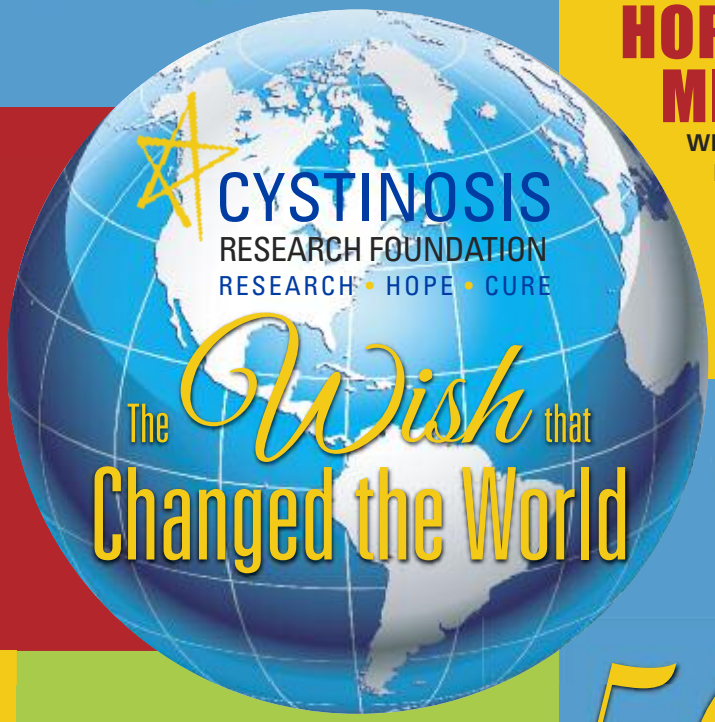

56
articles resulting from **CRF-funded** research have been **published**

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IN THE UNITED STATES WITH CYSTINOSIS
AND **2,000** **CYSTINOSIS PATIENTS THROUGHOUT THE WORLD**

Your generosity has funded
125 STUDIES
12 COUNTRIES

THE POWER OF
ONE


Thank you to everyone who has traveled with us on the journey towards a cure. Each of you has made a remarkable difference. We appreciate you more than words can express. Now, we hope you will stay with us to finish what we have so nobly started – ***to find the cure for cystinosis.***

DURING THE PAST
12 YEARS

WITH SUPPORT FROM ITS MANY FRIENDS, CRF HAS RAISED
$29.3 MILLION
TO BRING NATALIE'S WISH CLOSER TO REALITY