

# Cystinosis

MAGAZINE

FALL 2013

FOR FRIENDS AND SUPPORTERS OF THE CYSTINOSIS RESEARCH FOUNDATION



Team Hope

Currently, there is no cure for cystinosis, but

there is hope.....



**C**ystinosis 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 afflicts 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 affects 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 109 studies and fellowships in 11 countries. CRF has raised nearly \$22 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.

The Cystinosis Research Foundation is a non-profit, tax-exempt entity pursuant to Section 501(c)3. Federal Tax ID #32-0067668. 100 percent of the funds raised will support cystinosis research. All gifts are tax deductible.

*Photo taken at the 2013 Day of Hope Family Conference.*



Photo by Lars Wanberg

**CYSTINOSISRESEARCH.ORG**  
949.223.7610 • 18802 Bardeen Avenue, Irvine, CA 92612

# Table of Contents



6



5



16



12



8



49



18



26



44



34



47



43



56



23



40



52



32

A Letter from Nancy and Jeff Stack .....2  
 A Note from Natalie Stack .....5  
 A Journey of a Thousand Miles: Hadley Alexander.....6  
 Life is an Adventure with Landon and Jordan Hartz.....8  
 2014 Day of Hope .....11  
 Milestones: Olivia Little's Update .....12  
 The Good, The Bad & The Ugly: Dianne and Bruce Little ....15  
 My Amazing Little Brother Andrew Cunningham.....16  
 Keeping Hope at Full Throttle: Bailey DeDio .....18  
 Large Streams from Little Fountains Flow .....20  
 Thin as a Wafer, Big as Life: Dr. Acharya.....21  
 Zylar Rai Smethurst: A Little Princess with a Big Smile .....23  
 Dreamer. Fighter. Never Give Upper: Frieda Beilstein .....26  
 Donor Profile: Linda and Kris Elftmann .....31  
 Samuel and Lars Jenkins: Brothers Forever .....32  
 Supercalifragilisticexpialidocious: Tina Flerchinger .....34  
 Holt Grier: A Grandmother's Story .....40  
 Who Am I? Jennifer M. Loglisci.....43  
 Run with Mary Head .....44  
 Joshua Clarke: The Happy Trooper .....47  
 Sigma-Tau: Cystaran™ is Now Available .....48  
 Plates & Dishes: Gabbie Strauss .....49  
 Jenna and Patrick Partington Soar High Toward a Cure.....52  
 An Ongoing and Positive Journey: Hank Sturgis .....56  
 Library Science: Dr. Francesco Emma .....60  
 CCIR Looks for Advancements in Cystinosis .....61  
 Natalie's Wish 2014: Night of A Thousand Stars .....63  
 Sixth Annual Fore a Cure Golf Tournament .....64  
 Many Hands Make Lite Work.....68  
 It Takes a Village: Upcoming Events.....71  
 Cystinosis Patient Resources .....72  
 Matching Gifts Really Help.....74  
 Fourth International Cystinosis Research Symposium .....75  
 CRF in the News.....76  
 Raptor: Keeping Track of Cystine.....78  
 Lay Abstracts – Spring 2013 Grants .....79  
 Published Studies/Scientific Review Board.....82  
 2013 Call for Research Proposals.....82  
 2013 Grants Funded/2013 Interim CRF Grant Awards.....83  
 UCLA Research Study of Bone Marrow Transplant.....84  
 Cystinosis Research Foundation .....85



**On the cover:** Jake Krabe (left), seven years old, with his twin brother Austin. Jake was diagnosed with cystinosis when he was 18 months old.

**Please send your suggestions and comments regarding *Cystinosis Magazine* to [nstack@cystinosisresearch.org](mailto:nstack@cystinosisresearch.org). To receive our e-newsletter, *Star Facts*, send your email address to [zsolsby@cystinosisresearch.org](mailto:zsolsby@cystinosisresearch.org). The entire cost of *Cystinosis Magazine* is underwritten by friends of the Cystinosis Research Foundation.**



# Dear Friends and Family:

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

CRF was established in 2003 and since that time we have made astounding progress. We find ourselves reflecting on the early days of the foundation and we are amazed at how far we have come in just 10 years. When we look at where we are today, we are more hopeful than ever that we will fulfill Natalie's wish to "have my disease go away forever."

It is apparent why CRF has been so successful – we are supported by each and every one of you. We are a small orphan disease group with fewer than 2,000 people in the world, yet you have embraced our community and have surrounded us with your love, compassion and financial support.

## THE RACE

We have made tremendous progress funding cystinosis research but one thing has not changed – the life expectancy of a person with cystinosis is still 27 years. We are in a critical race against time. Cystinosis is a progressive disease that slowly destroys every organ in the body including the kidneys, eyes, liver, muscle and brain. As our children grow older, they deteriorate. The current medications prolong their lives but they are not a cure.

Although Natalie does not complain, we know she faces health challenges every day. She takes 52 pills a day to stay alive and healthy. The medications cause severe gastrointestinal side effects – there is no way to avoid them. Patients with cystinosis learn to live with the pain. As Natalie gets older she needs even more sleep for her body to recover, and more rest each day. Her energy level wanes the older she gets. Her eyes are covered with cystine crystals, which she tries to dissolve by taking hourly eye drops in an effort to save her vision. She lives in fear of going blind. We are always aware of the fragility of life and the urgent need to find the cure for cystinosis.

## A YEAR OF TRANSITION

This is a milestone year for us as parents – in December, Natalie will graduate from Georgetown University. She has applied to graduate school and plans to get a master's degree in social work. There are so many times that I catch my breath and ask myself where the years have gone. It seems like only yesterday that Natalie was starting her first day of kindergarten.

As parents of children with a terminal illness, we realize that time goes by too quickly. There are so many days that I plead with God to slow the hands of time. We need more time to watch Natalie grow and live her life, and fulfill her dreams before cystinosis advances.

It was 22 years ago when the doctor told us that Natalie had cystinosis and that she would likely die before she graduated from high school. Our beautiful baby was diagnosed with a horrific illness and we were devastated by the news. Not only were we scared and lost but we also searched for meaning and for hope. The life we had envisioned for our family was instantly transformed. We wondered if we would ever be happy again. Cystinosis changed the life path we were on.

## THE NEW NORMAL – RESILIENCE

The early years were filled with hospital stays, doctor appointments, a multitude of medications every six hours around the clock, changing sheets every night, ER visits and lots of tears, anxiety and worry. As time went on, we adjusted to our new life and found a “new normal.” What we initially thought would be impossible, became possible. What we were afraid of, we conquered. We had no choice but to face our fears and make each day count. We lived each day learning to celebrate the life we had. Cystinosis became our way of life – and our life before cystinosis became a distant memory.

Natalie's life is dominated by doctors, blood draws, clinical trials and medications. The challenges she faces as a result of cystinosis have become her strengths. Her fortitude, drive and courageousness have made her resilient. For our family, the darkest day of our lives – when Natalie was diagnosed with cystinosis – ended up being the start of a challenging new life filled with unexpected joy.

## CYSTINOSIS RESEARCH EXPANDS

Research is fueling our belief that we are closer than ever to reaching our goal of finding a cure for cystinosis. We continue to fund cutting-edge research and the research process continues to accelerate as a result of collaborations between our research teams across the globe. We have a synergistic team of scientists who are working every hour of the day to find a cure for cystinosis. We have made significant research advances, but there is more work to be done.

This year, CRF issued six new research grants during the spring application cycle totaling over \$1.6 million in new research grants. As a direct result of your generous donations, we have issued 109 research grants to scientists in 11 countries. Since 2003, the CRF has raised more than \$22 million for cystinosis research. Every dollar donated goes directly to research – 100 percent of the operating costs of the foundation are underwritten.



Photo by Geoffrey Regatz

## FDA APPROVAL

Our primary objective has always been to find better treatments for our children. CRF funded every bench and clinical research study that led to the discovery of a delayed-release form of medication. The original medication needed to be taken every six hours of every day but CRF-funded researchers at the University of California, San Diego discovered a medication that could be taken every 12 hours. We are proud to announce that the new drug was approved by the FDA on April 30, 2013 and is sold under the name Procysbi™. We accomplished this goal because of your generosity and commitment to our community.

## POTENTIAL NEW TREATMENTS

Our hope for the cure lies with the research being done in Dr. Stéphanie Cherqui's lab at UC San Diego. She is currently focused on autologous stem cell and gene therapy as a potential cure for cystinosis. Autologous stem cell treatment involves the patient's own stem cells which are harvested, genetically modified and reintroduced into the patient. Dr. Cherqui is working closely with the FDA to develop a clinical trial that we expect will be a reality within two to three years.

We are very excited to report progress in the area of corneal cystinosis. Dr. Jennifer Simpson of the University of California, Irvine, and Dr. Ghanashyam Acharya of Baylor College of Medicine in Houston have made great advances with their research. Corneal cystinosis causes eye pain, corneal scarring and potential loss of vision due to the buildup of cystine crystals on the cornea. Although cysteamine eye drops are available to reduce the crystals on the cornea, the drops must be taken every waking hour of every day to be effective. The eye drop protocol is so strenuous that it makes compliance all but impossible.

Dr. Simpson and Dr. Acharya have discovered a potential novel treatment for corneal cystinosis using nanotechnology. They are hopeful that there will be a clinical trial involving this new treatment within the next two years. We know you will be fascinated by the article about Dr. Acharya's work in this issue of *Cystinosis Magazine*.

## CYSTINOSIS RESEARCH HELPS OTHERS

Discoveries made by CRF-funded researchers and scientists are currently being applied to other more prevalent and well-known diseases and disorders. In fact, the delayed-release medication discovered by cystinosis researchers is now in clinical trials for Huntington's disease and NASH, a fatty liver disease caused by obesity.

CRF-funded stem cell research has the potential to help millions of people worldwide with other systemic diseases like cystinosis. Your donations have significantly impacted the cystinosis community and are now having a dynamic impact on others diagnosed with more common diseases. Because of you, hope for better treatments extends far beyond the cystinosis community.

## INSPIRING STORIES

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

We remain optimistic because you have been a source of strength for us. We are continually astounded by your generosity and your willingness to embrace our family and those who are affected by cystinosis. As we continue on our journey and our race against time, we are eternally grateful that you have joined us in our quest for the cure.

*With blessings from  
our family to yours  
Nancy and Jeff*

## *Dear Friends and Family:*

It's hard for me to believe that I am in my final weeks at Georgetown! I graduate in December with a bachelor's degree in psychology and a minor in French, and then return home to California. College has gone by so fast and I am so excited to move on to the next chapter in my life. After studying psychology for four years, I realize that I have a strong desire to help communities and individuals. My future plans involve getting a master's degree in social work in an effort to help others and change the world.

I recently applied to several social work master's degree programs throughout the United States. My first choice is USC, which has a top-rated program and I would be living in Southern California near my family.

This semester is different from all of the others. I am living off campus in an apartment, and I have an internship three days a week at the National Council for Adoption (NCFCA) in Alexandria, Virginia, where I am really enjoying my experience. NCFCA is a non-profit organization that advocates for adoption, and encourages and works with legislators on adoption-related issues. As a social work intern at NCFCA, I am learning about adoption policies, which allows me to advocate on behalf of children who need loving and permanent families. The staff and other interns are very genuine and compassionate people.

This is a time of transition for me. It seems like only yesterday that I made my wish to "have my disease go away forever," and I can't believe how much progress has been made through the years. I am so blessed to have such a supportive community of friends and families.

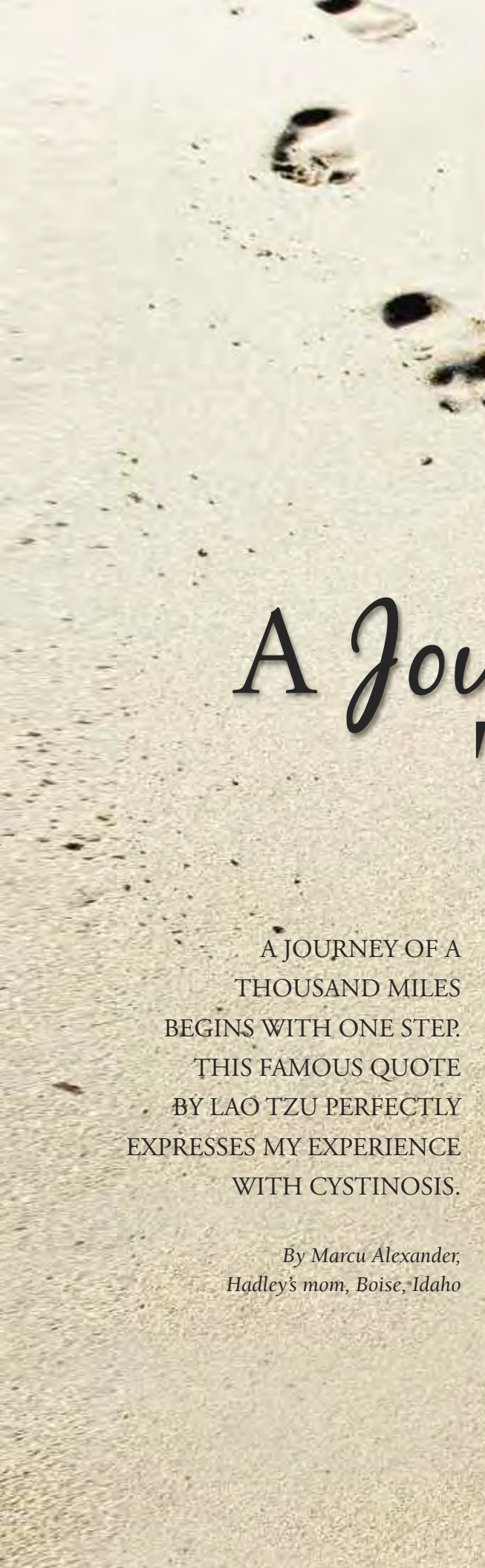
With your love and support, everyone in the cystinosis community will continue to have hope for the cure. Thank you for never giving up.

Love, Natalie



Photo by Geoffrey Ragatz

*Georgetown University  
Washington, DC*



# A Journey of a THOUSAND MILES

Hadley Alexander One Year Later

A JOURNEY OF A  
THOUSAND MILES  
BEGINS WITH ONE STEP.  
THIS FAMOUS QUOTE  
BY LAO TZU PERFECTLY  
EXPRESSES MY EXPERIENCE  
WITH CYSTINOSIS.

*By Marcu Alexander,  
Hadley's mom, Boise, Idaho*

*M*y family has spent the past year and a half on a journey that has changed our lives and taught us what is truly important. The first step on our journey began when Hadley was diagnosed. It felt like the world was slowly closing in on me and I wasn't sure I'd ever recover from the news. I was overwhelmed, full of fear and uneducated about the disease.

I spent several months diving in head first and learning as much as possible about cystinosis. Two weeks after the diagnosis, my husband, Hadley and I attended our first CRF Day of Hope Family Conference. We met wonderful people who welcomed us into our new cystinosis family with open arms. Many couldn't believe we made the decision to attend the conference so soon after receiving such difficult news. Ben and I never looked at it from that perspective. We saw it as a great opportunity to learn from the best and brightest doctors and researchers while connecting with families who are on the same journey.

Hadley has come so far since we learned she has cystinosis. She is a thriving, energetic and mischievous three-year-old. She recently began taking Procysbi™ and has handled the transition very well so far. We're all better rested and she likes watching the beads go through



the “tubie” into her tummy! Thanks to her G-tube, she tolerates all her medications well and rarely throws up anymore. As a result, she is eating better and growing! Since diagnosis she has gained more than ten pounds, grown several inches and her shoe-size has gone up five sizes.

She’s smart, funny and can hold her own in a crowd! Most importantly, she’s well-adjusted and happy! Her illness doesn’t affect her outlook or slow her down. In fact, she’s one of the toughest people I know. She doesn’t think about getting medication around the clock through her tubie, or visiting doctors several times a year. She doesn’t realize that drinking massive amounts of milk and water aren’t typical or that potty training is more challenging because of how often she needs to pee or the amount

she produces. All of these things are normal for Hadley and she thinks of herself as a big girl, just like her sister!

We have miles left to walk on this journey and only time will tell where it will take us. There will be bumps in the road and challenges along the way, but we are prepared for them and we will continue to enjoy each moment and do our best to stay positive.

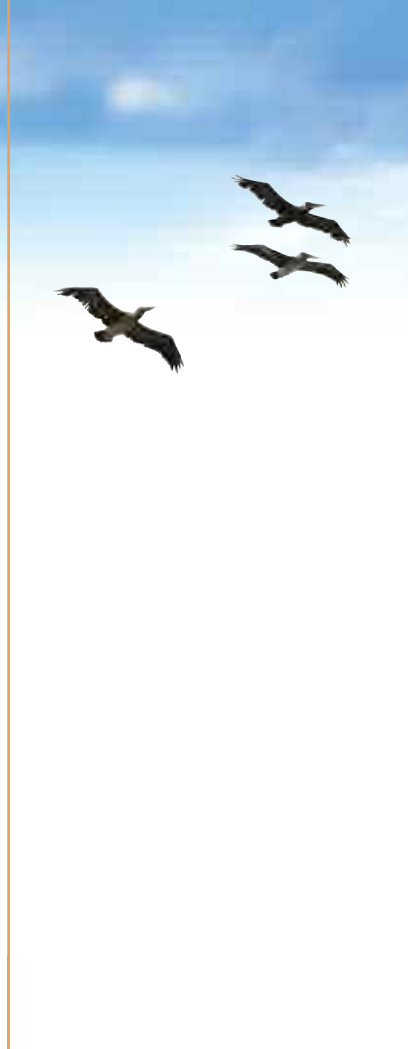
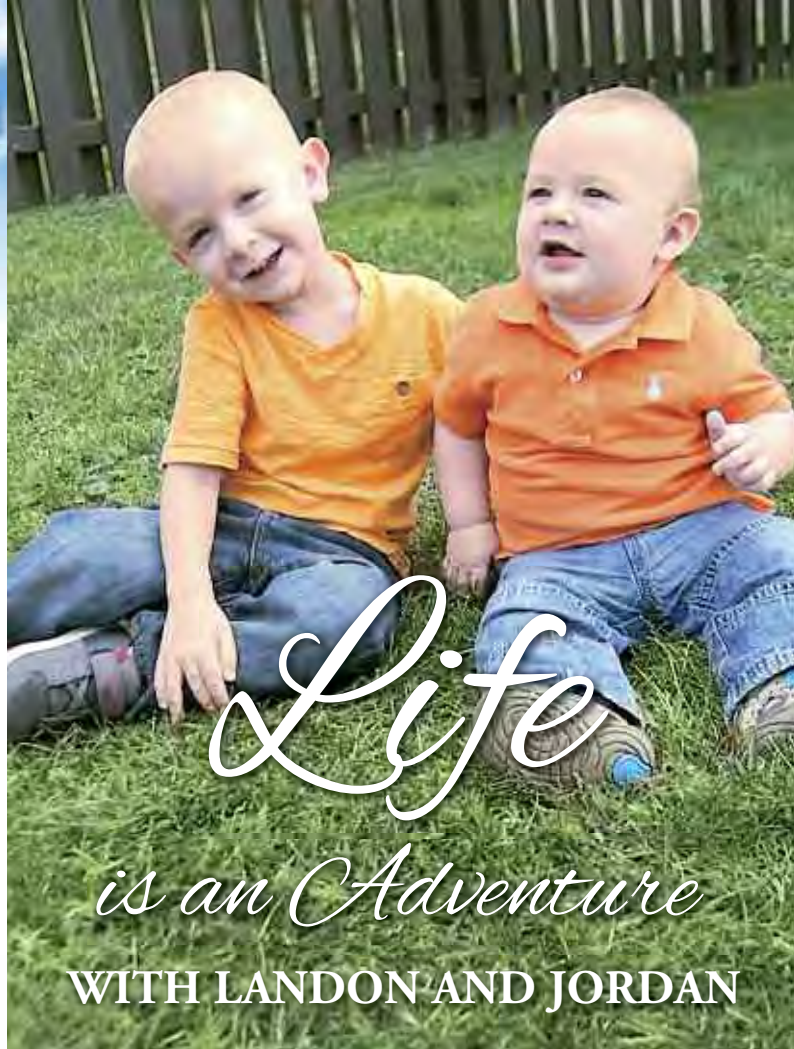
When times get hard or I start to feel overwhelmed, I remind myself how much worse things could be and I’m able to regain some perspective. If that doesn’t work, all I need to do is take one look at my sweet and spunky little girls and I’m reminded just how lucky I am!



*Hadley, right, with her sister Stella.*



Photo by Lars Wanberg



By Lauren Hartz, Landon and Jordan's mom, Pittsburgh, Pennsylvania

**M**ore than two years have passed since we sat in the hospital and learned that our perfect little boy was sick. We felt like we were in the middle of our worst nightmare. I remember telling the nephrologist that we did not want to know anything about the disease until we knew if Landon had it.

Curiosity got the best of me, and as I held back tears, I asked if Landon would live a “normal” life. Would he be able to go to school?

**LIFE WITH OUR TWO LITTLE BOYS IS AN ADVENTURE. NO ONE CAN GET JORDAN TO LAUGH QUITE LIKE LANDON CAN, AND THE BOYS TAKE TURNS TORMENTING EACH OTHER.**

Would he be able to play sports if he wants to? The doctor responded that, “Landon would live a normal life with some ups and downs.” He was right.

Landon has made tremendous progress in the last two years and really is a typical three-year-old. Our lives used to center on Landon's needs and his disease, but as we developed a new norm and a new

routine, we have had to think less and began to live and enjoy life more again. Two years ago, we had a baby who had a lot of anxiety, who was really behind on his gross motor skills, and had rickets from malnourishment. He vomited



daily, depended on a feeding tube for nutrition, did not stick a piece of food in his mouth, and was not anywhere near being on the growth chart.

Today, we have a little boy who goes to preschool and just today left his mom to go with a therapist he has never met. He runs, jumps, climbs and his bone health has dramatically improved. He rarely vomits and stays pretty healthy. He is finally on the growth chart for height and weight, and no longer uses his feeding tube. It was a very happy day when the nutritionist recommended that we stop the feeding tube, night feeds, and give him formula through a syringe. We have cut back on this formula because Landon eats pretty well. Originally, Landon did not eat at all, putting food in his mouth, pocketing it on one side before spitting it out. Slowly he began to put food in his mouth and swallow it. We couldn't wait until he would sit at the table and eat with us.

Landon has started to use Cystaran™ eyedrops, which were approved by the FDA, so obtaining them has not been a problem. When Landon was diagnosed, at 14 months, there were no crystals in his eyes. There were still none at 18 months, 2 years or 2 ½ years, but after his third birthday we learned there were crystals in his eyes, so within a month we started using eye drops. They are his least favorite medication but he cooperates with a bit of persuasion and persistence on mom and dad's part.

Jimmy and I have so much hope for Landon's future and for his health that we decided not to let cystinosis stop us from expanding our family. On January 15, 2013, we welcomed Jordan Patrick, our second little boy, to our family. Jordan was tested when he was two months old and we learned that he does not have cystinosis. We were relieved but sad. We grieved for Landon all over again.

Life with our two little boys is an adventure. No one can get Jordan to laugh quite like Landon and they take turns tormenting each other. Landon likes to lie on top of his brother and since Jordan is at the top of the growth chart and Landon, is at the bottom, he is usually not bothered by it but he gets back at Landon by pulling his hair. I have to admit that I worry about



Jordan getting bigger than Landon. I know it is coming and I don't look forward to it.

We remain committed to fundraising. Our annual events include a summer golf tournament, and a successful Halloween party in November. Research is crucial and supporting it, for us, is not an option. We had the opportunity to attend our first CRF Day of Hope Family Conference in April.

We were so proud to walk across the stage and present a check to the foundation for almost \$20,000 at the Natalie's Wish Celebration. Hearing the researchers speak and being around other cystinosis families motivated us even more and made us that much more hopeful about the future for Landon and others with the disease. We are excited for better treatments, and we can't wait for the day when we speak of the C word in past tense, "Landon had a disease called cystinosis but he was cured!"

**WE CAN'T WAIT FOR THE DAY  
WHEN WE SAY LANDON HAD  
A DISEASE CALLED CYSTINOSIS  
BUT HE WAS CURED!**



# LOTS OF LOVE FOR LANDON Second Annual Charity Golf Outing

Memories were made and \$16,000 raised for cystinosis research at the 2nd Annual Lots of Love for Landon Golf Tournament.

The tournament was held at the Ponderosa Golf Course in Hookstown, Pennsylvania on Friday, June 7, 2013.

Congratulations and thank you to Jimmy and Lauren Hartz – and to the organizing team including Landon’s uncles, Jason Whitfield and Jason Hartz, and friends Brad Hamilton and Chris Krasny – for their amazing efforts and dedication to help Landon and the cystinosis community.



A video montage of the day’s activities created by Lauren and Jimmy Hartz is available at: <http://animoto.com/play/E0g2gY92m7wXcwJg1zVDfw>

**Thursday, April 3 – Saturday, April 5, 2014**

Balboa Bay Club, Newport Beach, California

**The 2014 Day of Hope Family Conference will take place over three days and culminate on Saturday night at the Twelfth Annual Natalie's Wish Celebration.**

The conference will focus on the progress CRF-funded researchers have made over the past several years. Sessions will include:

- Research news about stem and gene therapy
- Novel eye treatment research progress
- Research updates related to kidney disease
- Update on the effects of muscle wasting
- Report on the Cure Cystinosis International Registry (CCIR)

All families are encouraged to participate in the discussions and question and answer sessions. Families will have a chance to meet, socialize and share their personal stories of life with cystinosis.

**More information to follow in early 2014, or contact Nancy Stack at [nstack@cystinosisresearch.org](mailto:nstack@cystinosisresearch.org) or Stacy Johnson at (949) 223-7610, [sjohnson@cystinosisresearch.org](mailto:sjohnson@cystinosisresearch.org).**



# Day of Hope

CYSTINOSIS RESEARCH FOUNDATION FAMILY CONFERENCE

## SCHEDULE

### Thursday, April 3

Check-in at the Balboa Bay Club  
Welcome reception and family dinner

### Friday, April 4

Conference sessions  
Followed by a family dinner celebration

### Saturday, April 5

Family brunch and social time

**Saturday evening** plan to join us at our Twelfth Annual Natalie's Wish Celebration.



Photo by Lars Wanberg



**Learn • Laugh • Share • Celebrate**



*By Erin Little, Olivia's mom  
Ontario, Canada*

**Our journey began on April 23, 2009: "We're pregnant."**

We were flooded with joy and instantly began loving our growing baby. For the next nine months we were consumed with preparing for our precious bundle of joy. We remember having conversations about who she would look like, places we would take her, college, what kind of temperament she would have, sports she would be involved in and most importantly how much we loved her. Having a "sick" child was never on that list.

# *Milestones*

## THE LITTLE'S UPDATE

It's funny how there are moments, situations and dates such as weddings, birthdays and anniversaries that just jump out in your mind. These dates will forever be remembered as key milestones in our journey of life. Well, our days aren't so typical.

**On Monday, December 14, 2009,** followed by an uncomplicated birth our beautiful and healthy baby girl, Olivia Ann Mae Little, weighing 7 pounds, 14 ounces and 20.5 inches long was born. It was the perfect day and we will never forget seeing her face for the first time. She was flawless. After two days in the hospital it was finally time to bring her home, we were so excited to begin showing her off to family and friends. After arriving home I will never forget the first thing we said to one another, "Where did all the adults go?" – and so our parenting adventure began.

**July 5, 2011 the phone rang.** “Mrs. Little we need you to bring Olivia to the hospital right away and just so you know you probably won’t be going back home today, so please bring what you think you may need.”

That morning we had been to our local hospital to have blood drawn to figure out what was going on. After spending the previous week in and out of the emergency room because of complications with Olivia’s breathing – without diagnosis – I was anxious to get to the hospital to finally get an answer. Assuming Olivia was suffering from anemia, I casually phoned Chad at work and told him we were heading to the hospital and I would keep in touch. We arrived at the hospital and Olivia was immediately whisked off to be poked and prodded while I sat with no answers. The next thing I knew, we were escorted to the helicopter and on our way to the nearest SickKids Hospital.

**July 12, 2011** “Mr. and Mrs. Little we are sending Olivia to see the optometrist to check for crystal buildup in her eyes. We want to rule out a couple of things,” the optimistic doctor said. I remember waiting in silence as we had no idea what to expect. We didn’t even know what they were looking for and we just hoped she didn’t have crystals. For being 18 months old Olivia cooperated so well and was happy to sit for the eye exam. After only seconds of looking into her beautiful blue eyes, the optometrist looked at us and said “Yup, she has crystals.” Our hearts sank. We had no idea what this meant but we knew it wasn’t the answer our doctor was hoping for. Without knowing what to do, what to think, or how to feel, we rushed back to talk with our doctor. It’s amazing how you become used to waiting – minutes felt like hours and hours felt like weeks. When the door finally opened, we received the devastating news.

We will never forget those words, “She has cystinosis.” We had no idea what it was and what it meant for our daughter. After listening to the doctor talk and show us diagrams – using words we have never heard – he ended the conversation with, “there is no cure.” We both quickly responded, “Not yet.”

began our new normal. Life picked up where we left off, working and taking care of Olivia. We wanted things to stay as normal as possible – our new normal just meant a new schedule 7 a.m., 1 p.m., 1 a.m. and 7 p.m. We were officially those people who always knew what time it was, as we awaited Olivia’s next



**July 23, 2011** We were finally back at home and as we walked in the front door, we had that feeling again, “Where did all the adults go?”

This time we came home with our precious daughter, loaded with bags of vitamins, syringes, gauze, vitamin schedules and a list of phone numbers in case of an emergency. Eager to be home, we unpacked and

dose of vitamins. During those months we kept to ourselves, trying to internalize what had happened and what it meant for our family’s future. When the time came, we reached out and that’s when things changed.

**April 19, 2012 – Cystinosis Research Foundation the Day of Hope Family Conference!** Until this day we carried on as normal or our



*Continued from previous page*

new normal: vitamins, water, doctors appointments, extra clothes, worrying about calorie intake, urine output, Poise pads and diapers, and all the other things that come along with the “new normal,” in the world of cystinosis. After the 2012 CRF Day of Hope Conference we carried on differently – it was our new, new normal. We no longer felt like we were alone in the battle, fighting for our daughter. We met people who inspired us to be strong and remain hopeful.

On the days when we feel like climbing to the top of a mountain and screaming, “We hate cystinosis,” we bite our tongues and remind ourselves to use that energy to make a difference.

After the Day of Hope weekend we returned home full of optimism, so much so, that we welcomed our second daughter, Harper Lynn, in May 2013. A week and a half after Harper’s birth, we found out that she did not have cystinosis, yet another phone call we will never forget. We were

overwhelmed with emotions – bitter sweet comes to mind.

Our hearts were filled with joy for Harper and ached for Olivia. We worry and dread the day that Olivia wants to know why she is unlike Harper. Although they are different in one way, they are alike in so many others – the most important being how much they are loved by their family. It’s probably not going to be the easiest journey but we will do it together and we believe the girls will be the best of friends along the way.

*To believe in life's gift is to keep hope burning bright.*

DIANNE LITTLE, AKA GRANDMA

**Where are we now?** Not a day goes by that we don’t count our blessings and that we are all here. We may not all be healthy but we are happy, we laugh, we cry, and Olivia throws in a good temper tantrum every once in a while to remind us that she’s just like every other three-year-old.

Olivia is joyful, which might even be an understatement. She is full of energy, kindness and zest for life. She adores her baby sister Harper and is always telling us to leave her alone so she can play and watch her sister. She is passionate about books and can’t get enough of them. She gives the best hugs, and she can’t wait until nap time is over so she can see her daddy because, as many people know, dads are way more fun than “rhythm and routine” moms.

We have our bad days and sometimes bad weeks but as Eleanor Roosevelt said, “Yesterday is history, tomorrow is a mystery, today is a gift, that’s why they call it the present.”

Like many other families, we are determined to help make a difference. Cystinosis is something we would like to archive in the history books. Our home is filled with confidence and we don’t take any day for granted.

As Oprah Winfrey says, “You get in life what you have the courage to ask for.” Our family has the courage to ask for a cure and one day Olivia and many others will have just that.

Thank you to the Stacks and all the doctors who dedicate their time and energy. We are forever grateful.





# THE GOOD, THE BAD & THE UGLY

By Dianne and Bruce Little, Olivia's Grandma and Pa, Ontario, Canada

## *The Good*

**We are blessed to have Nancy and Jeff Stack in our corner, fighting for a cure for cystinosis.**

There is nothing good about cystinosis, except that we have Nancy and Jeff, who have dedicated their lives to finding a cure for cystinosis.

When you look into Nancy's eyes, you see her pain for Natalie and for all the children of the "village." At last year's Natalie's Wish Celebration, as we listened to Jody Strauss deliver a speech honoring Nancy and Jeff for their work, we wondered how they keep up this pace day after day with such a positive outlook on life and for Natalie's future. How do you thank someone who creates a village – a family all wishing for the same thing and praying that it isn't too late for their loved one.

Natalie's wish has taken Nancy and Jeff on a journey for a cure, but also a fundraising crusade that inspires all who have become part of the family. Fortunately, that journey is almost over. Thanks to the research teams and doctors, it is only a matter of time before we find a cure.

## **THE BAD**

**A parent's health is as important as the child they are caring for.**

As we began the summer of 2011, Olivia fell ill on a day that none of us will ever forget. It was a day that would change Chad and Erin's life forever.

When your child is diagnosed with cystinosis, you go from nursing and raising your baby to a healthcare provider, on call 24/7. And it takes a toll on your family. Erin has the good fortune to be a stay at home mom, but I feel sorry for those less fortunate, who have to go to work knowing they have a child with cystinosis at home. If Olivia's diagnosis was God's plan he couldn't have picked better parents for her. And fortunately, Ontario has one of the best health care systems in the world.

Our advice to parents: Lack of sleep, stress and the day-to-day pressures of raising a family while being a round-the-clock caregiver can cause you to need doctor's care yourself. If you are unable to provide for your family it makes every problem worse. Look after yourself, keep your marriage alive and accept help! Your doctor can play a critical role in your mental and physical well-being.

## **THE UGLY**

**The Internet can be a dark place to turn for help.**

From the day Olivia was diagnosed, there were too many questions and not enough answers. Family members and friends turned to the Internet, which can be your best friend or your worst nightmare. You can't believe everything you read there. We heard so many predictions of what was going to happen to Olivia, and when it was going to happen.



Finally, Erin contacted the Cystinosis Research Foundation. She spoke to Nancy, who was able to calm her down by providing realistic advice. It was first-hand advice from a loving mother who has lived with this nightmare for 22 years. The CRF also helped Chad and Erin meet other cystinosis families, allowing them to vent and ask questions only a cystinosis family could answer.

Olivia is truly a "little" fighter, who continues to beat the odds that were stacked against her from the beginning, and she is up to the challenge regardless of what the Internet says.



*“I am a sister*  
**to my amazing little brother,**  
**Andrew”**

*By Kelsey Cunningham,  
Andrew’s 23-year-old sister  
Langdon, Alberta, Canada*

**I** will never forget that sinking feeling in my chest when I stepped off the school bus on a frozen January afternoon in 2005 and realized that something was different, and that something was wrong. My house lay darkened down the drive and the usual cars parked outside of the house were missing. I knew immediately that something had happened, and so I raced down the driveway as fast as I possibly could and threw myself through the front door. The only light in the house was coming from the answering machine. The red light flashed rhythmically like a lighthouse. A beacon of hope or a beacon of warning?

“Kelsey, your dad and I are at the hospital with your brother. Dad will be home as soon as he can to get you. Hang tight. Love you.”

I was 14, and I knew in that moment that my life would be forever changed. So began my story of being the sibling to a child diagnosed with cystinosis.



The next 13 days were spent in the ICU and J wing of the Alberta Children’s Hospital. My family was on a constant rotation, ensuring that my infant brother was comforted and loved, and that I was supported throughout that whole terrible time in my family’s history.

Seeing my brother hooked up to tubes and I.V.’s was traumatizing. Hearing him beg, “No thank you,” to the doctors and nurses drawing his blood work every three hours is an image that to this day breaks my heart and makes me want to cry.

incredible community of support. We would have never been given the opportunity to meet such inspiring people like those of you who attend the annual CRF conference. We would have never realized that the greatest gift we can give to others is lending our voice towards advocacy. We would have never understood the importance of family vacations. We would have taken for granted every single family activity we would have participated in. If it weren’t for Andrew’s diagnosis, I would not have chosen social work as my career path

and honest answer I can give to someone. I am who I am, because of my brother’s importance in my life.

My hope for my brother’s future is that he can achieve anything he sets his mind on, and anything that drives his passions. I want Andrew, who is now 10 years old, to be able to travel and see the world without having to bring a carry-on of medication with him. I want him to be able to attend any university he wants to go to without worrying if there is a specialist or doctor who will be able to help manage



## Living with Andrew then and now

Holding his frail little body while he balanced between life and death is something none of my family will ever forget. I think that any family living with cystinosis can relate to the utter helpless feeling you get when your child or sibling is in the fight of their life and there is little you can do to influence the outcome. Being 14 years old, watching Andrew struggle through his initial diagnosis made me feel as if I was drowning in helplessness.

For many families, this is the defining point of their young child’s life. For my family, however, this was the defining moment that made our family really and truly a “Family.”

If it weren’t for Andrew’s diagnosis, my family would have never become connected to such an

and I would not have decided to walk 100 km (62 miles) to raise awareness for kidney disease and organ donation. I would have never appreciated every single moment I get to spend with my brother.

A few days ago, I was in a job interview for a mentorship program when the coordinator asked me to describe myself.

The first words out of my mouth were, “I am a sister to my amazing little brother, Andrew.”

This is the answer I always give when asked who I am or what defines me. To me, this is the most important

his disease. I hope that he will be able to sleep through the night, every night, without having to worry about getting his meds on time. I hope he never has to have a transplant, or use eyedrops

to manage crystal build-up in his eyes. I hope he is able to taunt me, annoy me and make me laugh until we are room-mates in the old folk’s home.

Most importantly, I hope he is happy and healthy, and that he is never held back from chasing his dreams.

As for the future of his disease, I hope there isn’t one.

**My hope for my brother’s future is that he can achieve anything he sets his mind on, and anything that drives his passions.**



# Keeping hope at Full Throttle

## As Bailey waits for a kidney, the DeDio family spreads the word about donating life.

By Dennis Arp

**B**ailey DeDio is never happier than when he's riding his dirt bike. Wind whipping all around him, engine revving in his ears, dust from the desert trail billowing behind him – this is when he's totally alive and in his element.

These days, memories of such moments sustain Bailey, who turns 16 in November, at the same time that they now seem far from his grasp. As he copes with the debilitating effects of cystinosis, there are times when he struggles just to get out of bed.

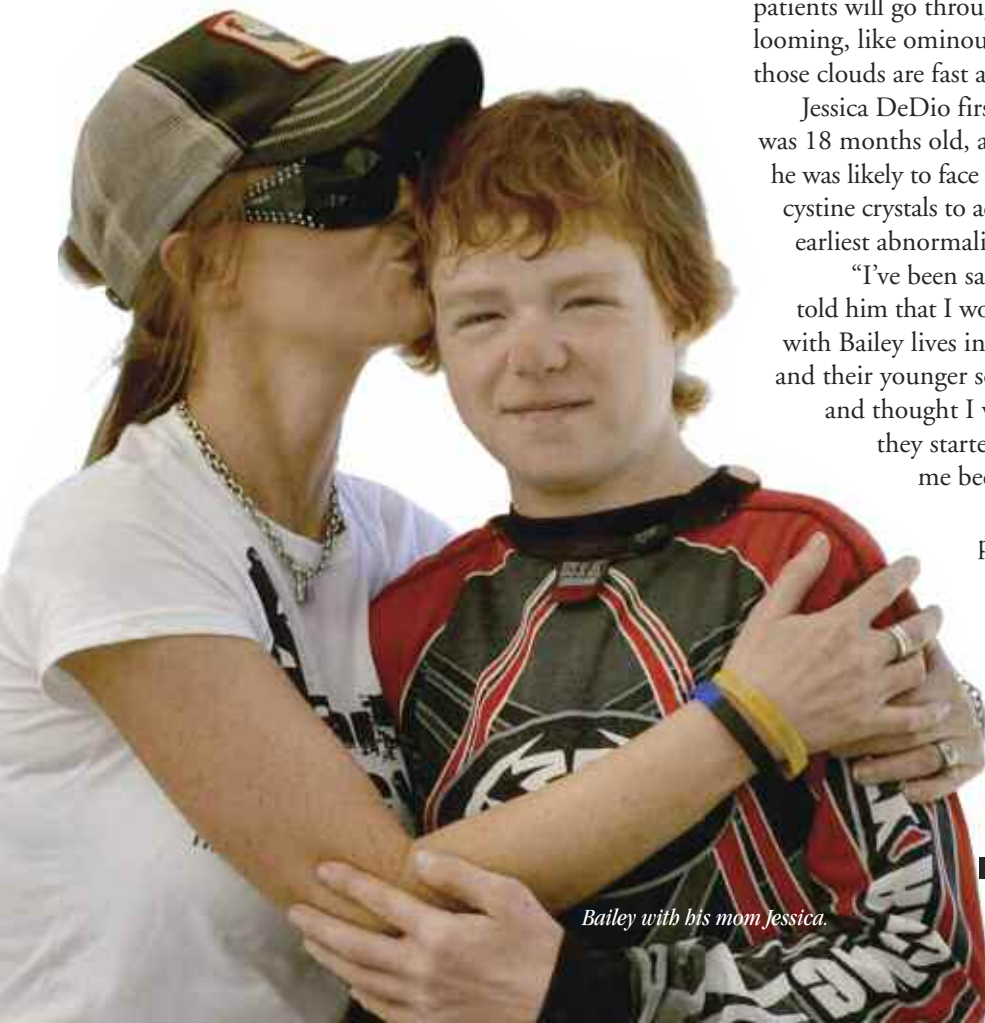
Bailey's kidneys are failing; they're functioning at about 15 percent of capacity, "and when that gets under 10 percent he'll have to go on dialysis," said Jessica DeDio, Bailey's mother. "He doesn't want to go on dialysis at all, and none of us want to see him hooked up to a machine."

Bailey and his family are coming to grips with difficult decisions and possibilities that are all too familiar to members of the cystinosis community. Because of advances in treatment, not all cystinosis patients will go through kidney failure, but that prospect is always looming, like ominous clouds on the horizon. For the DeDios, those clouds are fast approaching.

Jessica DeDio first learned that her son had cystinosis when he was 18 months old, and even then she heard from his doctors that he was likely to face kidney failure eventually. Cystinosis causes cystine crystals to accumulate in all organs of the body, but the earliest abnormalities are often seen in the kidneys.

"I've been saving my kidneys for Bailey, and I've always told him that I would donate to him," said Jessica, who along with Bailey lives in Fullerton, California, with her husband, Jay, and their younger son, Ryder. "I went through the whole process and thought I was on track to be a donor. Even at work, they started preparing to hire someone to replace me because it was such a no-brainer."

Then in June, "at the very end of the process, I found out that my anatomy wouldn't allow me to donate to anyone," she said. "It was an incredible shock to me, to Bailey and to our family. Honestly, that was one of the worst days of my life, because I had always promised him. That's what was supposed to happen."



Bailey with his mom Jessica.



*Bailey with his brother Ryder.*

The DeDios' journey has even more complications. Bailey's birth father is not in his life, eliminating another possible option for kidney donation. And Bailey has heard bad things about dialysis, fueling a strong fear of that course of treatment. Then, as if the family's cystinosis-related problems weren't enough, Ryder has developed serious health issues of his own.

But if the DeDios' story is about considerable challenges, it's also about resilience and the power of a supportive community. The family is not sitting back as Bailey sits on the donor wait

list. The DeDios are encouraging family, friends and everyone else to get tested as possible kidney donors.

Jessica's best friend, Tanya Chilcott, set up the Facebook page Bailey Believes – Donate Life “to help find a living kidney donor for Bailey as well as for other parents who are incompatible donors for their own children,” she says on the page.

“It's not only Bailey; so many other people need a kidney, why not donate to save a life?” Jessica said. “If we can spread the word – especially beyond cystinosis families, who have to save their kidneys for their own children – you never know what might come from this.”

This fall, DeDio family members and friends were also planning the second annual Bailey Believes Ride for a Cure, held Nov. 9. The event features a fun-filled day of dirt-bike riding and night of food and entertainment, including a silent auction and drawing with great items and getaways. Last year it raised \$14,000 for the Cystinosis Research Foundation. More information is at [www.cystinosisresearch.org/donate-for-bailey](http://www.cystinosisresearch.org/donate-for-bailey).

Such efforts do more than bolster Bailey's spirits as he copes with near-constant fatigue, as well as 75 pills a day, two shots a night and other cystinosis-related challenges. They ensure that hope remains a vital part of the family's equation.

“People's love and support keeps us going,” Jessica said. “I thank God every day for what I have. We're surrounded by positive people, and we all have faith that with the help of the CRF we'll find a cure.”

Meanwhile, amid the wait for a kidney, Bailey was planning to celebrate his 16th birthday with friends, family and the fixer-upper Toyota truck he bought with his own money, which he saved “his entire life,” Jessica said.

“When he has the energy, he works on it with my uncle and dad, and he washes it. He treasures that truck.”

For the treasured moments to come, the DeDio family's hopes are still revving.

**To learn more about donating a kidney, visit [www.kidney.org](http://www.kidney.org).**



*Bailey with his mom Jessica.*

## 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](http://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.

**[WWW.KIDNEY.ORG](http://WWW.KIDNEY.ORG)**



# Large streams from little fountains flow.

Cystinosis is one of 7,000 rare or “orphan” diseases in the United States. Collectively, rare diseases affect more than 30 million Americans. While there are only a small number of patients who suffer from any given “orphan” disease, knowledge discovered by studying one rare disease often leads to advancements in other diseases.

In fact, today CRF-funded research has the potential to help millions of people around the world with 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.

**Jennifer Simpson, MD, University of California, Irvine**

“Corneal cystinosis offers several advantages as a model from which **therapies for more prevalent eye diseases** will also benefit. Potential benefits include the use of stem cell transplantation and long-acting drug delivery systems. While both approaches are being developed for corneal cystinosis, they also have potential in other ophthalmic conditions including, corneal chemical burns, which results in corneal scarring; corneal surface dysfunction, which causes severe dry eye; and a number of conditions that require chronic drop therapy, most notably glaucoma and uveitis. **With the clinical endpoint of reduced crystals, corneal cystinosis provides scientific and regulatory validation for novel therapeutic approaches to these more common conditions.**”

**Daryl Okamura, MD,**  
University of Washington School of Medicine

“Chronic kidney disease (CKD) is a major health problem affecting 1 in 7 people in the United States that is quickly becoming a global health problem as well. Furthermore, kidney dysfunction is a recognized risk factor for poor outcome in a variety of disease states, yet there are few therapies to stop the progression of CKD and alleviate the oxidative stress within the kidney. Our recent study demonstrates that cysteamine bitartrate reduces oxidative stress within the damaged kidney and attenuates the progression of fibrosis by modulating the behavior of interstitial fibroblasts, a key cell in matrix production. **Our current studies are investigating the mechanisms of this effect with the hope of future clinical trials.**”

**Ranjan Dohil, MD, University of California, San Diego**

“Cysteamine has been available as a treatment of cystinosis for over two decades. But, it is only since the development of delayed-release cysteamine, with twice-daily ingestion and hopefully fewer side-effects, that interest in the use of this drug for other conditions has arisen. Delayed-release cysteamine is being evaluated in multi-center studies for non-alcoholic fatty liver and Huntington’s disease. **Recent studies also suggest that cysteamine may also play a therapeutic role in treating kidney fibrosis.**”

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

“Cystinosis is a metabolic disease that belongs to the big family of lysosomal storage disorders. These diseases often affect children and result in progressive multiple organ dysfunction and severe clinical complications. **Developing new therapies for cystinosis will provide insights to approaches that may have general applications for these diseases.**”

**Allison A. Eddy, MD, FRCP(C),**  
BC Children’s Hospital, Vancouver, BC, Canada

“In our recent CRF-funded research studies, designed to provide new insights into the question of why cysteamine has been so effective in preventing chronic kidney disease in patients with cystinosis, we discovered that it reduces kidney scarring – a process of kidney destruction that mediates all chronic kidney diseases. **If these findings are confirmed with further studies, they would provide rationale for testing the efficacy of cysteamine as a treatment for other forms of human kidney disease.**”

Cystinosis research now holds the potential to help millions worldwide.

# THIN AS A WAFER, BIG AS LIFE

By Dennis Arp

**Dr. Acharya's research breakthrough is tiny, but it holds the promise of continuous relief for corneal cystinosis patients.**

The wafer is so inconspicuous that when it's put into the eye, it barely registers. And yet it packs the enormous potential to change lives by the score. For now, that nanowafer is at the center of Dr. Ghanashyam Acharya's life as he works with passion to ensure that its transformative essence gets into the eyes of cystinosis patients as fast as human trials and the FDA allow. And to think that until March 2012, he didn't know anything about cystinosis and had no plans to pursue ocular medicine.

Dr. Acharya is an assistant professor at Baylor College of Medicine in Houston whose expertise is nanomedicine – the development of tiny drug-delivery systems that make treatment more efficient. He was invited to cystinosis research by Dr. Jennifer Simpson, a clinical professor of ophthalmology who is pioneering cystinosis treatments at the Gavin Herbert Eye Institute at the University of California, Irvine.

"I got a phone call from Jennifer, who told me about her research into corneal cystinosis and the need for a sustained-release delivery system," Dr. Acharya said. "She asked me to help, and I immediately said, 'I'll do it.'"

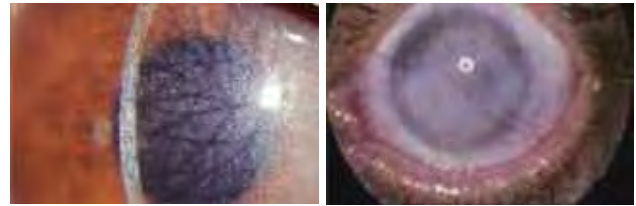
That's because he immediately understood the depth of the need. He learned that cystinosis causes a painful build-up of cystine crystals in the eyes and on the cornea – a feeling like always having sand in your eyes. Cysteamine drops provide relief, but the treatment has to be repeated every waking hour, so compliance is low.

"There was a need for a whole new technology – a nanowafer drug delivery system," Dr. Acharya said. "This had never before been used in the eye."

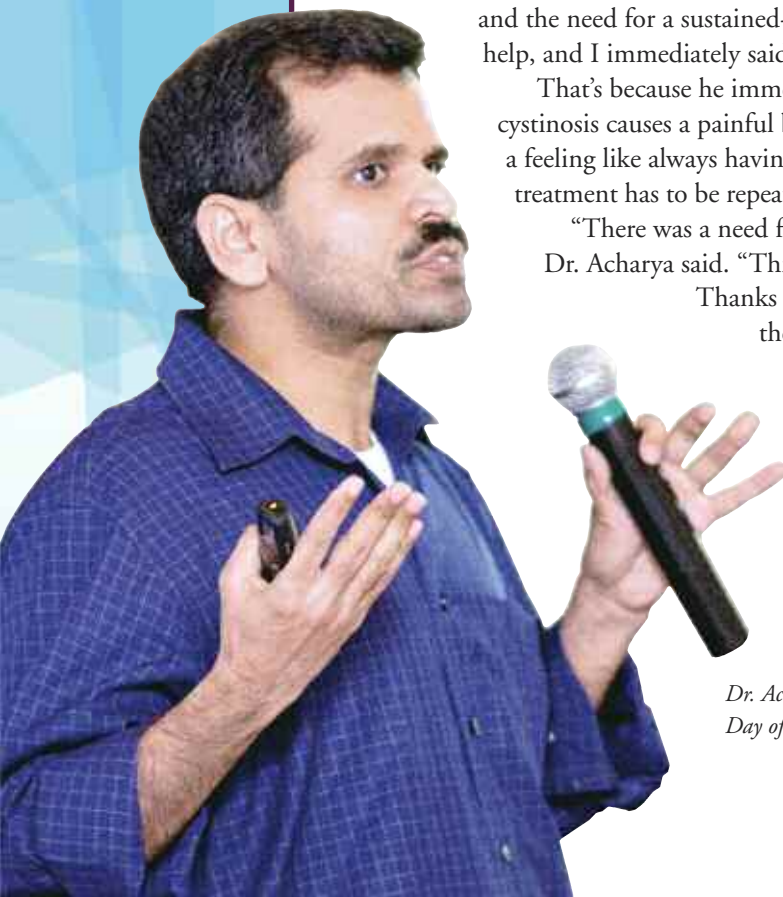
Thanks to a grant from the Cystinosis Research Foundation (CRF), the doctor was able to load nanowafers with cysteamine, which is released into the eye as the wafer dissolves. The hope is that a single wafer will clear away crystals for up to a week.

"It's working fantastic in the mouse model," Dr. Acharya said.

"This nanotechnology holds the promise of revolutionizing treatment," said Nancy Stack, CRF chair. "This is by far the most exciting project we're funding at the moment because it offers new hope for a better treatment."



*Photos show crystal buildup on the cornea, which causes cystinosis patients extreme photophobia, great pain, and possible loss of sight.*



*Dr. Acharya at the 2013 CRF Day of Hope Family Conference*

*Continued on next page*

Mouse-model testing has been so effective that human clinical trials are expected to begin next year at three sites: UC Irvine, Texas Medical Center in Houston and a site to be determined.

“The beauty is that the nanowafer material and the drug already have been approved (by the Food and Drug Administration),” Dr. Acharya said. “It’s just the combination of the two that needs to be tested for efficacy. That’s why I’m very optimistic.”

The process still includes many variables, so Dr. Acharya is reluctant to speculate on when approval might come. But he knows that it can’t come soon enough for cystinosis patients and their families.

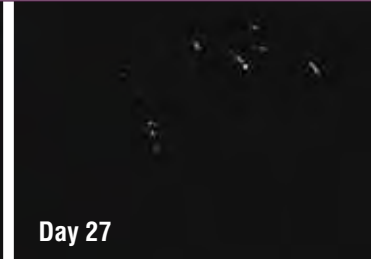
The urgency hit home in April when Dr. Acharya attended the CRF Day of Hope Family Conference in Newport Beach, California. “The conference was an eye-opener. At the end of the first day, I was emotionally drained from meeting the patients and the parents. To see such small children, 4- and 5-year-olds, wearing dark glasses and drinking so much water. I realize that time is the most important thing here. We need a treatment as soon as tomorrow,” he said.

“Ultimately the experience was incredibly positive,” Dr. Acharya added. “It bolstered me to work with even more intensity. As a scientist, I’m quite a happy person in the lab. In many ways it’s a dream world, a highly protected world. The conference put me in touch with reality.”

Dr. Acharya, the scientific researcher, and Dr. Simpson, the practicing physician, work thousands of miles apart, but they make a remarkable team.



Day 1



Day 27

*Efficacy of the nanowafer in dissolving corneal crystals in cystinosis knockout mice.*



*Dr. Acharya in his lab at Baylor College of Medicine in Houston*

Department of Defense to develop nanodrug delivery systems to treat eye injuries suffered by soldiers during combat. And he’s in an early stage of development on a transdermal-patch drug-delivery system for treating cystinosis patients.

The more Dr. Acharya extends the reach of his research, the more he is drawn back to the connections that foster such breakthroughs.

“People like Jennifer Simpson encourage me to do things that I have never done before,” he said. “We know exactly what we want to do, and that’s why I’m optimistic that we will get there together.”

**Corneal cystinosis research has become the purpose for my life, which is what I told Jennifer when we met face to face for the first time at the conference.** DR. ACHARYA

“I develop the tools, and Jennifer uses them to improve patients’ lives on a day-to-day basis. This is where a multi-disciplinary collaboration comes to fruition,” Dr. Acharya said.

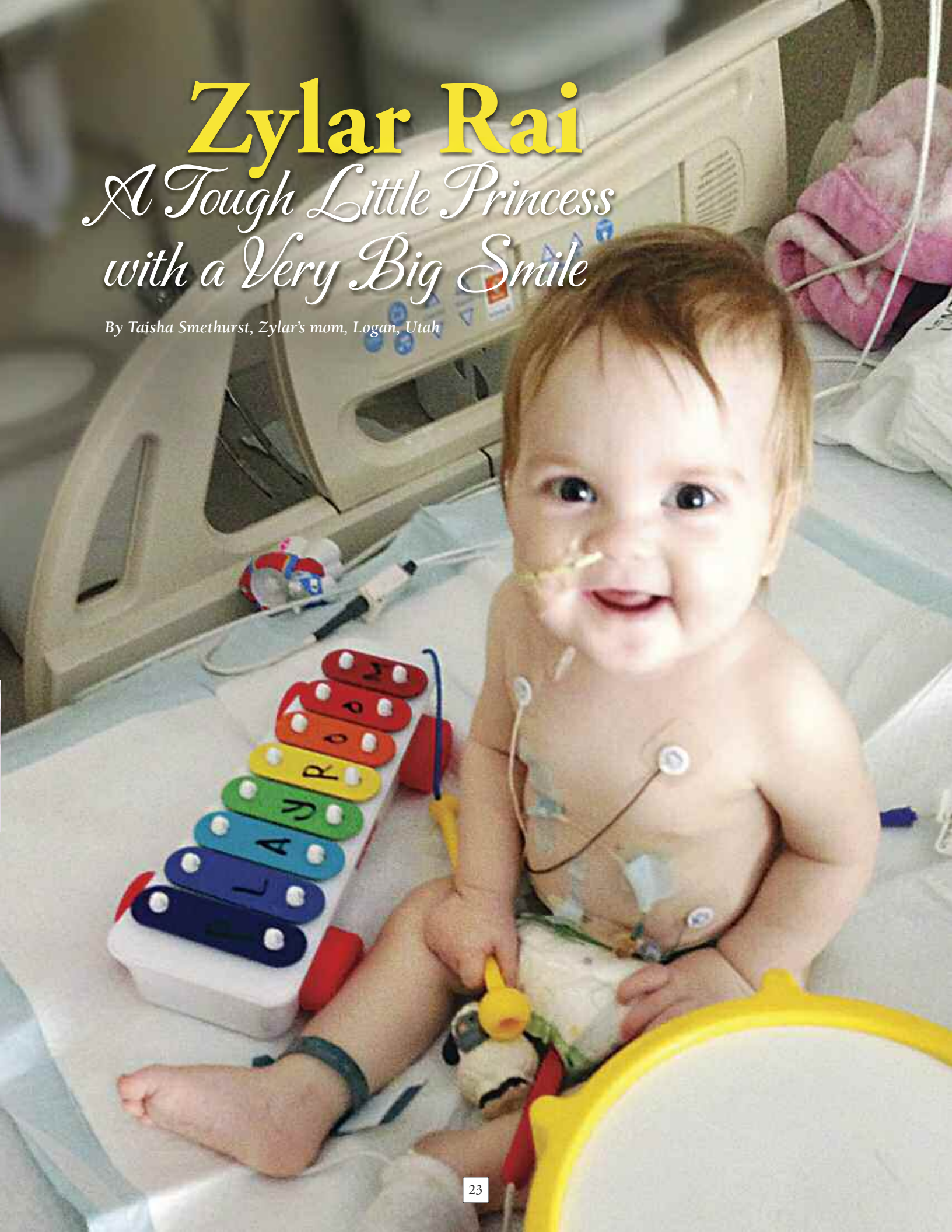
The fruits of his research are branching into other areas as well. Dr. Acharya and Dr. Stephen Pflugfelder have recently earned a \$2.1 million grant from the U.S.

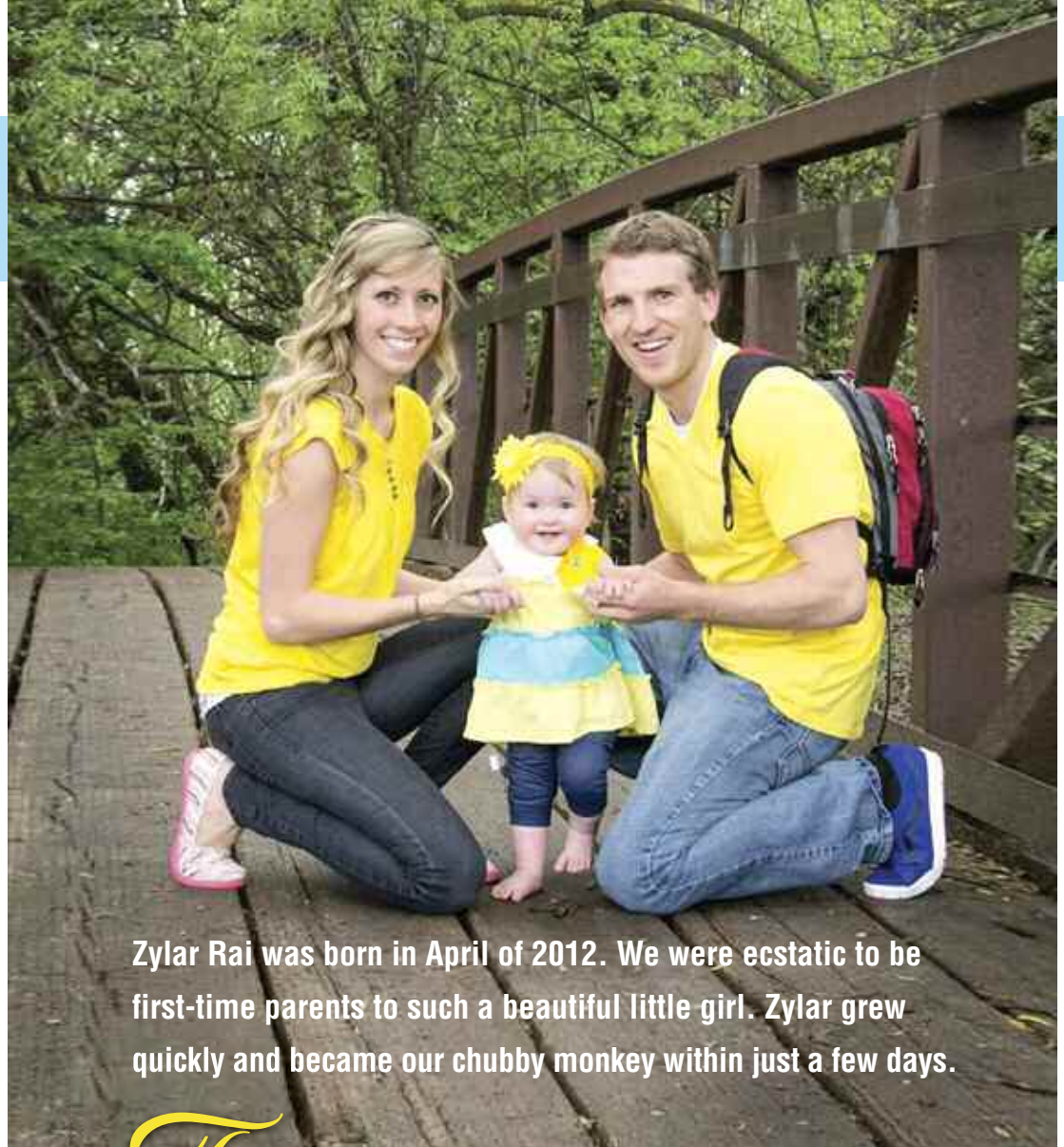


# Zylar Rai

*A Tough Little Princess  
with a Very Big Smile*

By Taisha Smethurst, Zylar's mom, Logan, Utah





**Zylar Rai was born in April of 2012. We were ecstatic to be first-time parents to such a beautiful little girl. Zylar grew quickly and became our chubby monkey within just a few days.**

**F**or the first six months of her life Zylar was always in the 97 percentile for her size, but at only three months, she had her first overnight stay at the hospital. She was admitted for pneumonia, treated for dehydration and sent home the next morning. Zylar got sick often and easily but was always a trooper, giggling and playing all of the time.

By Christmas of 2012, we started to suspect something was wrong with Zylar. At eight months of age we were constantly calling her doctor, taking her in for appointments, and going to the local InstaCare. Time after time we were told that it was flu season, babies get sick a lot and that we were just

being over-cautious, first-time parents. At about that time, Zylar also started to refuse to eat anything. She had always been a good nurser but when we introduced food at about six months of age she refused to eat. After a few months, she began to throw up if anything got in her mouth.

Doctors and even close acquaintances would tell us that she would eat when she got hungry enough. And then, Zylar began to refuse to nurse, as well. We were literally watching our baby starve to death while others told us she was just stubborn. As she quickly lost weight, people offered excuses like she's moving around more and is just losing baby fat.

In one month, Zylar dropped from the 97 percentile into the 50s, and she went from sleeping through the night to sleeping only in half-hour intervals and crying when she was awake. She started to want only water but after chugging it she would throw it up moments later.

In February of 2013, Zylar was admitted to our local hospital through the ER for an overnight stay. She was admitted for Respiratory Syncytial Virus (RSV) but was only treated for dehydration and extremely low potassium. The next morning we went home. After three more long weeks of watching our daughter deteriorate in front of our eyes,

we reluctantly took her to the InstaCare again, knowing we would just be turned away as we had been a number of times before.

However, that afternoon, a new, young doctor we hadn't seen before took notice of Zylar. As soon as he saw our very lethargic, fragile little girl, he knew something was wrong. He held her against his shoulder for only a few seconds before he was out in the hall making phone calls. We were quickly admitted to the local hospital (again), but this time on orders from the doctor to take it slow and really determine the underlying problem.

On March 7, we began our long journey of diagnosing and treating Zylar. We spent a hectic night at the local hospital placing IVs, running tests, taking x-rays, getting ultrasounds, and having labs taken as frequently as possible for Zylar's little body.

At some point during the night our pediatrician suspected Fanconi

Syndrome and called a renal specialist at Primary Children's Medical Center. We were told that first thing in the morning Zylar would be put on Life Flight to Salt Lake City's Primary Children's Medical Center.

The weather that morning was inclement and the helicopter couldn't make it into our small valley. A medical plane was then scheduled to pick up Zylar at the small local airport, only to be canceled soon

after because of weather. Finally, the decision was made to put Zylar in an ambulance to take her through the canyon to meet the helicopter at a hospital

on the other side.

Because of the hectic circumstances, Zylar's dad and I were not allowed to ride with her. After a VERY long two-hour drive, we were finally reunited with our sweet baby. To make matters worse, the paramedics were her first official "babysitters" since we had always taken Zylar everywhere with us.

We were extremely blessed because the renal doctor who saw Zylar that day at Primary Children had dealt with cystinosis before. He took one look at her charts and knew that what he saw was the same disease he had seen while working with other patients. After checking her eyes and discovering the crystals, he was certain Zylar had cystinosis.

On March 8, 2013, Zylar was unofficially diagnosed with cystinosis. Lab tests returned from California confirmed the diagnosis two weeks later. Had we gone to Primary Children's during any other

week – when a doctor who had never seen or treated cystinosis was on duty – our diagnosis may not have been so quick or simple.

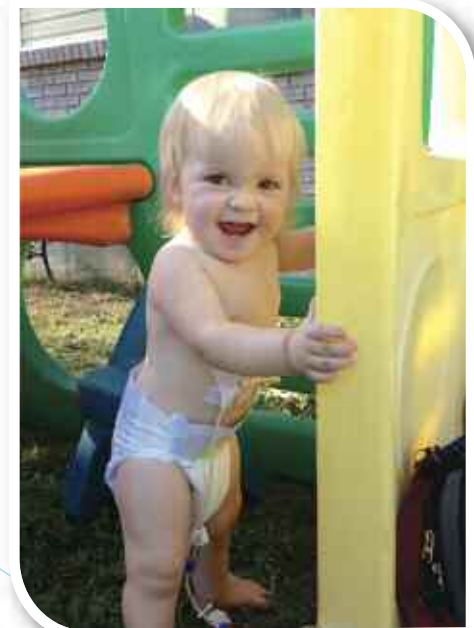
We spent five days in the Pediatric Intensive Care Unit before we were moved to the infant unit, where we spent the next three and a half weeks stabilizing Zylar. Those weeks were not without some very scary incidences when all we could do was watch and pray for our baby. She also had a G-tube placed during our stay.

After a month in the hospital we were finally able to come home. At first, it was intimidating having 24-hour feeds and so many medicines, but soon it became part of our everyday life.

Today, six months after diagnosis, Zylar is growing and gaining weight. She still has rough days full of tears and vomit, but our tough princess pulls through with a very big smile.

Since Zylar's diagnosis, we have made some amazing friends in the cystinosis community and have been inspired by many parents, children and others with the disease.

**Zylar still has rough days full of tears and vomit, but our tough princess pulls through with a very big smile.**



**Dreamer.  
Fighter.  
Never  
Give Upper.**





**My name is Frieda Beilstein. I am 23 years young. I live in Hawkesville, a small town in Ontario, Canada. I am blessed with a wonderful family and friends.**

**I** have one younger brother and his name is R.J. He has always been my number one supporter and my absolute favourite person.

R.J. can always make me laugh even when I don't want to smile. Family means everything to me.

I enjoy travelling, kayaking, going to the beach, and being outside. I love exploring the world and trying new things (as long as I can sleep in first). I like reading and I believe learning is a never-ending journey. I am studying education and hope to be a teacher someday.

This spring I plan to graduate from Conestoga College with a diploma in early childhood education. I believe in following your heart, helping others and being the best person you can be. I feel obstacles are put in your life to make you stronger, and I believe in never giving up. The biggest obstacle in my life is cystinosis.

I was diagnosed with cystinosis when I was six months old. I became very ill; my skin was yellow, I lost weight and was extremely dehydrated. After a series of blood work and other tests, I was diagnosed with this very rare genetic disease.

Immediately, I was transferred to nephrology specialists, and admitted

to the hospital where I started taking numerous medications. While my diagnosis was a shock to my family, "cystinosis" was not a foreign word to us. My mother's cousin, Judy, had cystinosis. Being familiar with the disease and knowing the battles I would likely face in my life was scary for my family. Sadly, around the time I turned one year old, Judy passed away. She will always be loved and missed by many.

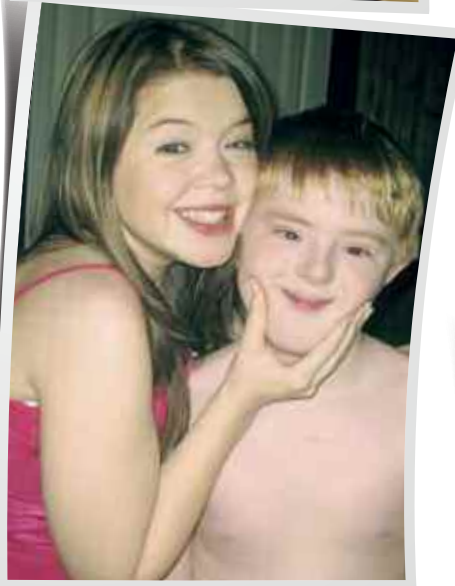
Stories about Judy are very special to me. Precious memories of her are like secrets being whispered in my ear.

I always imagined if given the chance we would have been best friends. I imagine us side by side always supporting each other's fight against cystinosis. And even though I cannot see her, I know she is watching over me. I wish there was as much research and education available when Judy was alive as there is today. Cystinosis care has surely come a long way since the 90s.

I think most of you reading this will agree when I say that living with cystinosis becomes normal faster than expected. It's not fair when medications,

*Continued on next page*





doctor's appointments, and emergency hospital stays fill your daily routine. But that's exactly what living with cystinosis is.

When people ask me questions about cystinosis it sometimes upsets me because I've had cystinosis for as long as I can remember and there's really no way to explain something that is so normal to me. It's like explaining breathing. Some symptoms are visually obvious: height (I'm 4'8" AND A HALF ... the half is extremely important), pale features, malnourished, etc. The internal symptoms are usually what hurt me the most, and the worst part is that no one sees

them and they are impossible to explain. Some days I feel so sick, but for the life of me, I can't explain where I feel ill.

In my early years, the worst part of living with cystinosis was probably the eye drops and taking all my medications. I was in and out of the hospital. I quickly became immune to getting blood work, as I bravely surrendered my arm for tests.

I had several types of feeding tubes before we finally found one that didn't fall out every time I coughed. That was the G-tube. My mother and grandma tell me stories of how difficult that was. The worst

part was changing the G-tube. I remember the nurses holding me down and pulling the tube out of my stomach and replacing it once a year. It was a painful procedure, but I needed the tube for my feedings and medications.

My mom says I was always so brave and did everything the nurses and doctors asked with a smile no matter how much pain was involved. My grandma remembers how often I would vomit. "People sitting across from you would get hit," she laughs when we reminisce. I was spoiled with love from my family. They

would always risk a cuddle with me even knowing they would most likely get vomit on them or urine from my always-full diaper.

I also had rickets in my bones from the kidney disease, so I had to be more careful when playing, but I think I had a pretty normal childhood.

I didn't realize I was different from my friends until my preteen years. It was weird because my friends always wanted to be skinny, and I was excited when I gained weight.

several awards as well. It was a great achievement for me.

Unfortunately, I was not able to attend grade nine with my classmates because my dialysis time increased to 16 hours a night and I just wasn't well enough. So for that year I had dialysis, some home schooling, and completed my 40 volunteer hours. I was still positive and I did the best I could. I had a lot of support from family and friends. They always made sure I had a smile on my face.

keep up and stay healthy. I wasn't in any classes with people born the same year I was. I still didn't have a lot of energy and my sick days piled up.

And of course, I realized I was even more different. I just wanted to be like everyone else and I didn't want to take medication all the time. The cystinosis started to take a toll on my nerves and emotions. I began to lose interest in life, I felt like a failure, and I lost hope. I was diagnosed with major depression and a generalized anxiety disorder, which wasn't surprising to my doctors, but it was hard for me to accept. I like being independent, and with another health problem it was getting more difficult.

Fortunately, soon after, I started at Conestoga College where I began to thrive. I took leadership courses, and really challenged myself to be more independent. I worked with a campus counsellor on "who I was" and what I wanted in life. I was even able to live on campus. I felt very blessed that I was able to work through my struggles and obstacles.

I realize that I am still different. I still get exhausted but I have learned to cope with cystinosis. I've learned how to balance my energy and enhance my skills. And others accept

*Continued on next page*

**Everyone can give something: time, money, talent, anything. I strongly believe when you give as much as you can and then some more, God always gives back to you.**

I stopped growing taller and discovered I was the shortest in every class.

My classmates were involved in sports. They all could run faster than me, and I lost in every physical contest. Gym was not my best subject. I had a lot less energy than the other children. And I questioned why the others in my class never had doctor appointments in big cities or why they never had to stay in the hospital.

However, what bothered me most was that I never won the award for fewest missed days of school. I had a lot of sick days no matter how hard I tried to have perfect attendance.

In 2004, my kidneys were in complete failure. I spent a lot of time in the hospital that year. I was put on peritoneal dialysis and the transplant waiting list. With this type of dialysis I was able to have treatments at home while I slept at night. Luckily, I was also able to balance school. Not only did I graduate from elementary school with my class, I managed to earn

In June 2005, I was blessed with a kidney transplant and I started feeling much better. I had different medications and my appearance changed but I was happy and ready to get back to my social life.

High school was a lot harder than I expected, probably the hardest part of my life so far. My kidney worked great but I still had cystinosis and its symptoms. I still struggled to





me for who I am, and most important, I've learned to accept myself. I may not be able to do everything my friends do, but there are a lot of things I can do. College was great for me. I was on the Dean's Honour list, I was involved in extracurricular activities, and I was a Resident Advisor for a whole year.

Unfortunately, I am currently on sick leave because in January 2013 I rejected my kidney transplant and I'm back on dialysis. But it's okay – I am optimistic about my future. I am determined to accomplish my dreams.

I have an amazing family and friends. They are so supportive and back me up in everything I do. My mother planned a fundraiser for

CRF's Canadian partner, Cystinosis Awareness & Research Effort (CARE) and for awareness for organ donation. Both causes are very close to my heart. I am so thankful for all the work my mom did and to everyone who helped. The event was an amazing success. We raised \$24,794.22, and we had a lot of fun. We had speeches (even my paediatric nephrologists came to speak about cystinosis), a live auction, dinner and dance. When I stood on stage and looked into the crowd, I was amazed that so many people had come to support me.

It is so touching to read all the stories in this magazine, and to see all the organizations supporting research. I've always believed in giving to causes

that are close to your heart, so it is inspiring to see how many people have joined this community of support. Everyone can give something: time, money, talent, anything. I strongly believe when you give as much as you can and then some more, God always gives back to you.

I want to continue to be involved in fundraising for cystinosis, and other connected organizations. A few weeks ago I participated in the 5km walk for The Kidney Foundation. I amazed myself when I crossed the finish line – *I did it!*

Sometimes the future – and knowing all the science about cystinosis – scares me. I don't know how sick I might get. Right now it's difficult to get out of bed, and I feel very weak. I'm not as independent as I wished I could be. And after a busy day with family and friends – fundraising, kayaking, any sort of day outings – it takes me about a week to recover.

But there is always hope and I know God has great things planned for me. My obstacles may be bigger than most people's but I'll still make it in my own time because I'm dreamer, a fighter, and a never give upper.

**I wish the best to all of you and your families.**

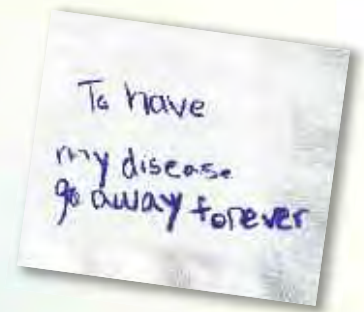
**Stay beautiful,  
Frieda**





# A Moment That Changed Their Lives

By Dennis Arp



**From the moment 10 years ago when Linda and Kris Elftmann first heard about Natalie's Wish, a little girl's dream became their inspiration.**

The Elftmanns are owners of Noelle Marketing Group in Orange County, California, and in 2003 they were enlisted to help create invitations, the program and other supporting materials for the second Natalie's Wish fundraising event. It was during a first meeting with Nancy Stack, Cystinosis Research Foundation (CRF) chair, that the Elftmanns learned about the restaurant napkin on which Natalie Stack wrote her now-famous 12th-birthday wish – “to have my disease go away forever.”

“It's really the wish heard 'round the world,” said Kris Elftmann, president of Noelle Marketing Group and now a member of the CRF Board of Trustees. “This courageous little girl, now a remarkable young woman, was able to capture in a few words not just the dream shared by everyone who has cystinosis but also the challenge for all of us who embrace the cause of finding a cure.”

As is their norm, the Elftmanns pursued with gusto the effort to promote the Natalie's Wish fundraiser, which the year before had been highly successful but on a smaller scale.

“You don't have to know Nancy very long to know that she's both brilliant and a big thinker,” Kris said. “She and Jeff had organized a cocktail reception, and they raised an incredible amount, \$100,000. Together we saw the opportunity to double or triple that. We said, ‘Let's go for it.’”

The second event attracted 250 guests and raised almost half a million dollars, with all of the money going directly to critical research, as it still does today.

“I remember that night vividly,” Kris said. “We sat at a table with a dad and his son, and with one of the earliest and most important researchers. Linda and I have been around

many charitable events and been involved in philanthropy for a long time, but we've never been to an event where you could literally hand the check to the researcher to keep the research going. That left an enormous impact.

“It's as direct as philanthropy can get. Here is the donor, here is the doctor, and here is the child. There's no giant staff, no overhead. And it's still like that today.”

The money is critical, of course – without it there would never have been the monumental progress achieved over the past decade, nor the breakthroughs that continue today in labs on multiple continents. But the human connection is also vital, Linda Elftmann said.

“I don't remember specifically who spoke at that first event, but I remember that it was quite an emotional evening,” she said. “It was our first chance to meet the children and their families – to learn what they go through and how brave they are, yet they don't let it get them down.”

The Elftmanns have been passionately involved with the CRF ever since, contributing their time, talents and treasure. They are honored to be part of the team that produces *Cystinosis Magazine*.

And they have helped the Natalie's Wish Celebration become one of the most successful events of its kind, last year raising \$2.2 million for research that impacts not just cystinosis but other rare diseases.

“This cause has captured our hearts,” Linda said.

“We've been part of many wonderful organizations,” Kris added, “but we've never been on a ride like this. A few families started out wanting to help a single little girl, and now you feel like the work of the past 10 years could change healthcare worldwide. It's astounding.”

And the ride continues.



**HOW DOES ONE GO ABOUT RAISING  
MONEY FOR CYSTINOSIS RESEARCH?**

**WE HAVE BEEN SO IMPRESSED AND INSPIRED BY  
THE MANY FAMILIES AND COMMUNITIES WHO  
HAVE RAISED MONEY TO CURE THIS DISEASE.**

**WE WANTED TO HELP TOO, BUT  
WEREN'T SURE WHERE TO START.**

Our first splash into fundraising was a letter campaign to our family and friends. We sent out letters to about 100 people, and posted that same letter on our blog and Facebook pages. We were overwhelmed by the generosity of our donors. In 2012, people sent us \$9,626. We were ecstatic and so grateful to make a contribution to the Cystinosis Research Foundation, which works tirelessly to find better treatments and a cure.

We went to the CRF Day of Hope in April 2013 (first time for Stephen and Lars), and we came home infected with more enthusiasm. We believe wholeheartedly in the cure, and we decided that fundraising would always be an important goal for our family.



# Samuel and Lars

## BROTHERS FOREVER

*By Ashton and Stephen Jenkins, Samuel and Lars' parents, Salt Lake City, Utah*

**B**ut where to go from here? Most days we are just happy to keep our heads above water. After the Day of Hope Conference we settled back into our daily routines, so organizing another fundraiser seemed like it might be the straw that breaks our camel's back.

Enter Mary Ann Franson, a friend of Ashton's family. She is from the neighborhood and church congregation in which Ashton grew up. She deals with her own health issues, but after our letter-writing campaign, she was galvanized to action. She organized a massive yard sale in Provo, Utah and recruited her five children, their spouses, and other members of the congregation to donate and gather quality furniture and other items. Mary Ann planned every detail, so when the day arrived, everything ran perfectly. Students from Brigham Young University gave up their Saturday morning to unload a U-Haul filled with furniture and other goods. A group of young girls baked and sold cookies and cupcakes, raising more than \$100. By day's end, the yard sale raised an amazing \$3,328.

We are so grateful to Mary Ann and her family for their selfless love and willingness to help. It's people like them who give us the courage and hope to deal with cystinosis every day.

Samuel and Lars are doing great. Samuel has been on Procsybi™ for two years now, having joined the phase IV trial in November 2011. We recently received word that our insurance will cover the cost of the drug, so we will be making our last bittersweet visit to Stanford for the trial.

We are so grateful to have been a part of the study, and we will miss meeting with Dr. Paul Grimm. We are grateful to Xiaoxiao, Suvarna and Allison, the study coordinators we have been privileged to work with.

Sam just started pre-school and loves it. At the ripe old age of four he has finally got the hang of this potty-training thing, and he can go all day without a diaper. We still haven't figured out how to keep him dry at night. He is used to a regimented schedule and often reminds us when it's time for a bolus feed or medication dose. He takes the Cystaran™ eyedrops eight times a day without complaints, and his ophthalmologist was astonished by the improvement in his crystal burden at his last visit. He still gets tube feeds

for the bulk of his nutrition, but he is doing much better at eating. He loves nuts, cheese, chicken, fish and cheeseburgers. He is all boy and loves duplos, trains, swords, baseball and singing "I'll Make a Man out of You," (from Disney's *Mulan*) at the top of his lungs.

Lars is a chubby and charming wunderkind. You should see this kid eat. He grabs food off our plates if we aren't fast enough. There are few foods he won't eat, probably because he's tasted the sickening sulfur of Cystagon® since he was one month old. He continues to tolerate Cystagon® well, although the faces he makes when we squirt it in his mouth are both hilarious and heartbreaking. So far we've only had to add carnitine to his medication regimen, in addition to Prilosec



Photo by Lars Wanberg

*Ashton, Lars, Sam and Stephen Jenkins at Stanford Medical Center.*

and vitamin D. To our amazement, his renal function panels remain normal. We have been fortunate to avoid the growth delays, polyuria, electrolyte losses and endless vomiting that plagued Sam.

Lars wants to do everything that Sam does, and he's thick as thieves with Macey, our family dog, the beneficiary of Lars's poor table manners. He loves balls, cars, juice, climbing, walking and iPads. We can't wait to get him on Procsybi™.

**Thank you to the Cystinosis Research Foundation and all the generous people that support this great work. We are amazed by the tenacity and boldness with which they move forward toward a goal we can all get behind.**



Supercalifragilisticexpialidocious!  
Even though the sound of it  
Is something quite atrocious  
If you say it loud enough  
You'll always sound precocious

# SUPERCALIFRAGIL

By Denice Flerchinger, Tina's mom, Clarkston, Washington

The last few months have been a welcome change. Summer was spent getting Nichole packed and off to college, I miss her terribly.

Catherine is a high school senior and preparing for college next fall, and Tina is going to miss her partner in crime.

Both Nichole and Catherine have had such a positive influence on Tina's life – cheering her on in times of accomplishment, and encouraging her to forge ahead in times of challenge.

All this talk of college has me anxious about Tina's future. The typical cystinosis patient takes a dramatic decline in health at the age of puberty. I am fearful of what Tina's future will be. Every day I have to renew my trust in God otherwise these thoughts consume me.

Mark and I are extremely proud of our daughters – all three are beautiful, joyful, vibrant souls with much to offer this world.

Tina is preparing for a piano recital in which she will be playing the familiar piece titled "Supercalifragilisticexpialidocious" – it is the longest word in the dictionary and defined *extra ordinary*. This fits Tina's personality to a "T." Even in the face of adversity she always puts on a big smile and makes the best of every situation – *extra ordinary* indeed!

Tina is the typical fourth grader – she loves to ride her scooter, play with friends, and all things hot pink. Her days are spent dodging chores and playing with her dog, Sam. When asked, she describes herself as funny and nice. I would add independent and feisty to that.

Photo by Lars Wanberg



# ISTICEXPIALIDOCIOUS



To know her is to love her. She is a delightful little gal that can carry a tune, and her giggle will melt your heart. The only thing that distinguishes Tina from her peers is the nine medications she takes each day, in the form of 48 pills. The new

school year has brought a reminder of the stringent schedule Tina must follow to stay healthy. Her day starts at 6:30 a.m. with a can of nutrition, 19 pills, and several sets of medicated eye drops to keep her from going blind.

Her daily regimen ends at 9 p.m. Night times can be challenging; she has an unquenchable thirst due to Fanconi Syndrome, which is why she needs to urinate every 30 minutes. Tina has never slept through the night, had a sleepover, or slept in. However, school is going well. She loves the social aspect of it and has found a new passion, reading.

In August, after three years, Mark and Tina made the twenty-fourth and final trip to Stanford Medical Center – bittersweet indeed. Her new medication has been life-changing. I cannot comprehend how we survived six years of G-tube feedings, 2 a.m. medications, and around the clock nausea and vomiting. Tina has been through much in her short life.

We are eternally grateful to the Cystinosis Research Foundation for funding the research and development of

the new medication. Together, with CRF's help, we are all changing the history of cystinosis.

This year has been tough for many – several newly diagnosed and a handful awaiting kidney transplants, some as young as eight. When Tina was diagnosed, in 2005, we were told she would not live to see double digits. Last month she turned 10 years old. Every day is a celebration of her precious life. We know Tina's organs are slowly deteriorating, but we continue to pray that she will remain strong until a new treatment is available.

As Thanksgiving approaches, my heart is filled with thanks for each of you – whether a researcher or a donor your support is making Natalie's wish, Tina's wish and the wish of all these children and adults living with cystinosis a reality. We are on our way to a cure!

*A spoonful of sugar doesn't always help the medicine go down. We may have retired most of Tina's oral syringes, but the deluge of medicine goes on. This is a one-week supply of her new medications. She was excited until she saw how many pills there are – 48 a day. She's such a trooper, not one complaint.*





By Holly Bonnalie,  
Tina's cousin  
Lewiston, Idaho

*A Small*

*To Help Tina*

A FEW MONTHS HAVE PASSED SINCE THE 4TH ANNUAL TINA'S HOPE FOR A CURE EVENT TOOK PLACE AT ROGER'S TOYOTA SCION IN LEWISTON, IDAHO. AFTER AN EVENT OF THIS MAGNITUDE, IT TAKES BOARD MEMBERS AND THE FLERCHINGER FAMILY SEVERAL WEEKS TO RECUPERATE AND CATCH UP ON OTHER FACETS OF OUR LIVES.



# Town Comes Up Big

*A*s for me, one week to the day after the event, my husband and I welcomed our first baby, a delightful little boy named Hudson. He is a second cousin to Tina, and she has already been able to hold him several times. The hot days of summer have allowed each of us to reflect upon the wonderful evening of love and support the Flerchingers and the entire cystinosis community received on that May night.

Without question, it is always a little bittersweet to see another year go by. In one sense, it is exciting to know that we are funding research dollar by dollar. Yet, each year we hold on to that glimmer of optimism that this will be our last time planning the event, and that Tina and all those who suffer with cystinosis will be completely healed.

We have come so far from where we started. I clearly remember the day my Aunt Denice mentioned it was time to plan a fundraiser, and asked if I would consider being on the board.

When Tina was diagnosed, no one in our small community understood what a cystinosis diagnosis entailed. It was a foreign word to everyone, with people constantly looking at us with quizzical expressions on their faces, saying, "What is the name of that disease again?"

*Continued on next page*



*Photo above: Tina with cousin Holly Bonnalie and her new son Hudson. Holly is a board member of Tina's Hope for a Cure, and one of the driving forces behind the success of Wine, Stein and Dine.*

*Photo left: Tina, with her dad Mark, drawing raffle winners at the recent Wine, Stein and Dine.*



*Tina with the ladies of Tina's Hope for a Cure Board – (L to R) Mary Ausman, Tina, Sherry Seeh and Holly Bonnalie.*



*(L to R) Friends – Judy & Dr. Mike Rooney, Kerri & Rob Sandaine, and Mary Tatko.*



*(L to R) Check-in – Chris Kessler and Karol McBride.*

Over the past five years, it has become a familiar term to those around us. It's known as the "Baader-Meinhof Phenomenon:" once you hear a new word or concept, you hear it again and again. People now know what cystinosis is, and its effects on the human body. They understand that it has to be cured – there is no other option.

This is where our entire community has stepped up with great force. Nearly 300 people attended this year's event, with guests from all walks of life and outlying towns arriving to rally around this cause, this journey so very close to our hearts, and this beloved, brave little girl. Businesses and individuals donated a wide variety of auction items, allowing people to bid on unique offerings ranging from vacations to magnum-sized bottles of wine. Guests sampled multiple ethnic foods, while they sipped on locally handcrafted wine and beer.



**JOIN US FOR WINE,  
STEIN AND DINE 2014  
ON SATURDAY, MAY 17**



Bernice Schweiter, mother of one of our board members, Sherry Seeh, passed away January 24, 2013 at the age of 91. Bernice and her deceased husband Walter enjoyed sitting near the Flerchingers at church and watching the girls grow. It was through her parents that Sherry and her husband Jack were introduced to the Flerchinger family 10 years ago.

Only a few months after Tina was born, Bernice was concerned about Tina's health and her frailty. When Tina was diagnosed with cystinosis she didn't know what that meant for Tina's future, but she thought about Tina and her family and prayed fervently for them.

When Sherry joined the Flerchingers in their newly formed non-profit, Tina's Hope for a Cure, Bernice was interested in what research was being done in cystinosis and read all the information she received from the CRF. She cheered Sherry and the group on in each year of their fundraising and came to all the events (except in 2012 when she was too frail to attend).

After Bernice's passing, her friends were honored to make memorial donations in her name to Tina's Hope for a Cure. Memorial donations in Bernice's name totaled \$1,350. We know she is smiling down on Tina and the Flerchingers and happy to be a part of fundraising for cystinosis.





*The Flerchinger family with the Rogers family, which has become a tremendous supporter of Tina's Hope for a Cure and cystinosis research.*

**The entire event raised an astounding \$115,000!**

Our annual *Fund a Cure* was wildly successful, with individuals raising their paddles to donate more than \$56,000. If you had looked around the room during this portion of the evening, you would have seen women and men alike brushing tears from their cheeks. Perhaps this is the most uplifting and exciting portion of the event – the portion where we can see first-hand money going directly towards research.

I wholeheartedly believe that people's generosity is directly related to the encouraging news we constantly hear about new cystinosis treatments, stem cell research, and the ultimate knowledge and belief that we will find a cure. It seems that the cure is just around the corner, inches within our grasp, so close we can envision our lives without cystinosis being a term we discuss on a daily basis. We never know when that one dollar raised will be the exact amount needed to put cystinosis into the history section of medical textbooks, so we will continue to fundraise until our dream becomes reality.



*Tina visits with her sisters and cousins.*



*Rich and Ryan Rogers*

## We Couldn't Do It Without Them!

Rich and Ryan Rogers give new meaning to the words – we couldn't do it without them – when it comes to Tina's Hope for a Cure. Rich and Ryan are the dynamic father-son team that owns and operates Rogers Toyota-Scion in Lewiston, Idaho. They stepped forward three years ago when they heard about Tina's annual *Wine, Stein and Dine* to donate their showroom as a venue for the event. Things haven't been the same ever since.

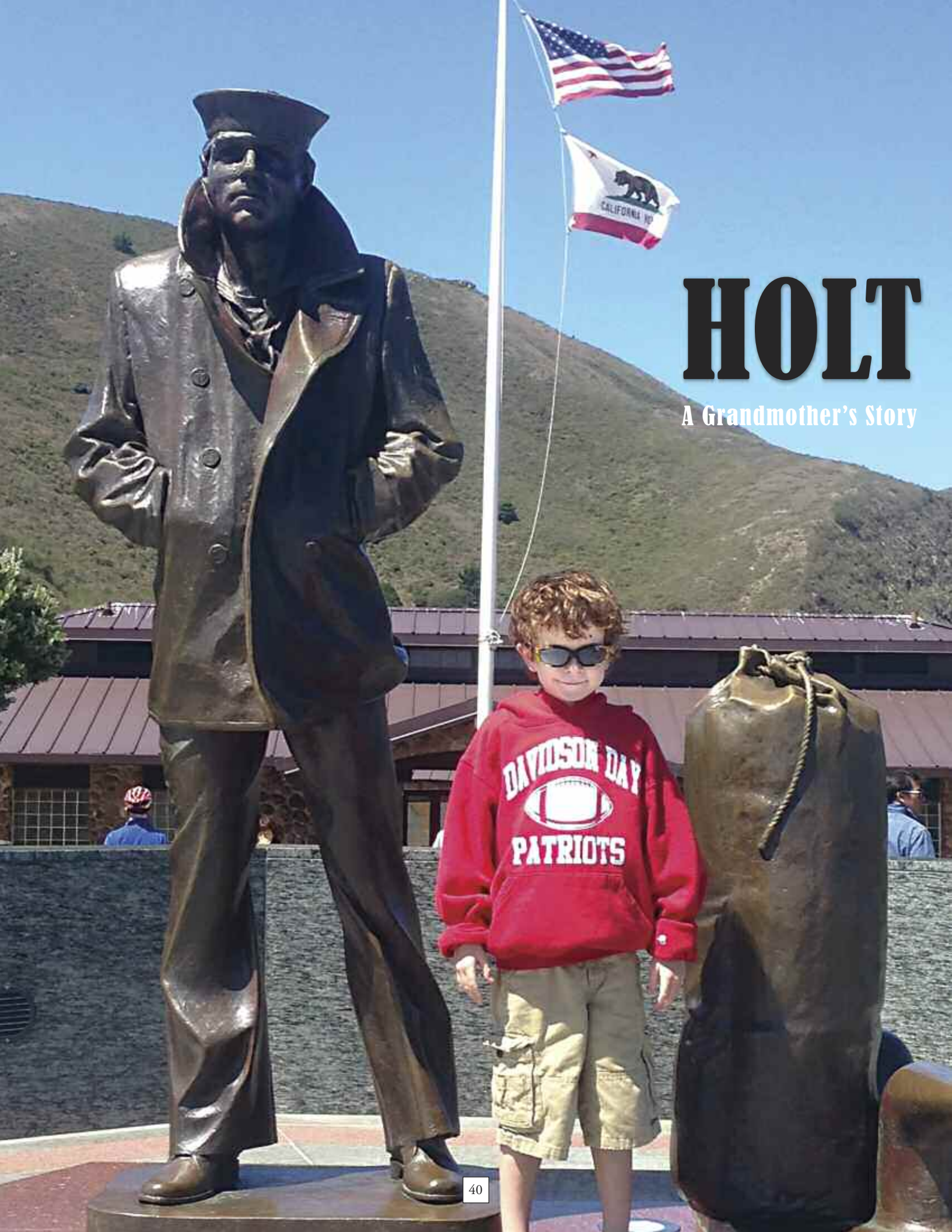
The event, now held in the spacious, jewel-like showroom-turned-hotel-ballroom, has become the most anticipated event in their small community. And with the Rogers' help, the event has grown faster and more successful than anyone could ever have imagined.

The Rogers' staff almost literally rolls out the red carpet when they see event volunteers drive in to set up the event. They also roll up their shirt sleeves to help in any and every way possible to make the event a huge success. ***And they do it all starting the day before the event.***

This year, like each of the previous years we've used their facility, the Rogers Toyota staff and Rogers families went even further (if that's entirely possible). They donated \$5,000 to Tina's Hope for a Cure.

There is almost no way to calculate the support this exceptional family has provided to Tina and others with cystinosis. They are certainly wonderful friends and community members. They're also truly remarkable role models to each of us, and we cannot thank them enough for all they do as we continue on the quest to find a cure for cystinosis.





# HOLT

A Grandmother's Story

**My son, Jason Grier (and Holt's father), asked me to write about being the grandparent of a child with cystinosis. I am humbled and honored by his request. I hope that other grandparents, especially those whose journey with cystinosis has just begun, will take a small amount of comfort knowing that we all start out, after learning of our grandchild's diagnosis, with the same feelings of anguish, frustration and helplessness.**

# GRIER

By Carole Bradley, Holt's Grandmother

**W**hen the pundits and politicians talk about our country's future, they often refer to the huge national debt with which our grandchildren will be saddled. Prior to March of 2007, I may have been more concerned with that issue than I am now. Why? My grandson, Holt Grier, was diagnosed with cystinosis in 2007. Now, the future means finding a cure for cystinosis so that Holt and others who suffer with this disease will have a future.

What must it be like, as a parent, to hear the diagnosis confirmed via a phone call from your child's nephrologist, that your child has a disease for which there is no cure? Holt was 16 months old when his mother, Chrissy, took that call.

I happened to be at Chrissy and Jason's home the day that call came. She took the call in the next room, while Jason and I waited for her to return and give us the results of the diagnostic testing that Holt had undergone. When she returned with the news, she was crying softly and was holding a piece of paper on which she had written notes taken during her conversation with the physician. She shared the devastating news that Holt had cystinosis, a metabolic disease, which attacks the body's organs – and that there is no cure.

The ears hear, but the mind cannot accept this diagnosis. *What? How can this be?* This child came into the world happy and healthy. So did I notice that by the age of 10 months he wasn't walking, like his siblings had? Had I noticed that he was very thin and pale? Had I noticed that he had an insatiable thirst and consumed liquids so rapidly that I thought the tops of his sippy cups must be leaking? Yes, I did notice these things, but since I knew that Holt had regular visits with his

pediatrician, I told myself that they were on it. All would be well.

In my heart, I knew that this sweet boy was not thriving. When I saw him, I was engaged in an emotional battle of denial between my heart and my brain. My heart told my brain that Holt would gain weight, start walking and be healthy. Yet, should I mention my concerns to Jason and Chrissy? I kept quiet. Holt's health would improve ... it had to. Nothing else was acceptable.

*Continued on next page*



*Carole Bradley  
with Holt*



## Hope for Holt

*Continued from previous page*



Without research, there is no cure.

I could write many pages about the toll this disease takes on those who suffer from it and on those who love them. I would like to share one incident that I will never forget: it took place one pre-dawn morning in Jason and Chrissy's kitchen, not long after Holt's diagnosis. I had gone down to make coffee and found my mountain-of-a-man son leaning over the sink, his broad shoulders heaving up and down, as he quietly

sobbed, in that dark room. I believe that he was overcome with the helplessness that he felt at not being able to make his son all better. I felt that helplessness ... I had no words.

**Positive talk time!** We are so grateful for the improved treatments from those days when our loved ones had to be given their meds every six hours. We now have real hope for a cure,

as the dedicated people work so diligently on behalf of the cystinosis community.

We are blessed beyond measure to have Nancy and Jeff Stack, and their daughter Natalie involved in our search for the cure. Natalie's long-ago

wish to have her disease go away forever was the genesis for the growth of the Cystinosis Research Foundation. The CRF and our donors ... they keep Natalie's wish and our hope alive.

There are so many diseases out there, and all need to be eradicated. I speak about cystinosis because it is one of the meanest and it affects one of the dearest people I know, my almost 8-year-old grandson, Holt. The scourge of cystinosis affects the youngest of our loved ones, before they have ever had a chance to enjoy good health. So we must never, ever give up on funding that research. Keep doing what it takes to find the cure and make it available to every patient. Have no limits when seeking donors to support the research. It is a continuous process until the cure is found.

*Love in the search for the cure,  
Carole Bradley*

**N**ow we know the diagnosis, no more denial. Our family and friends are on the Internet, trying to learn all we can about this disease, which none of us had ever heard of. Much of the information was very clinical and not easy to understand for lay people. I yearned for just one tiny article, or even a sentence that said a cure was imminent.

No such article appeared and so, we did what all families who are blessed with strength and commitment, and who love and cherish their children do: we mobilized and began to plan ways to raise money to help fund the research for better treatments and the cure. That was what we had to do.

**Keep doing what it takes to find the cure and make it available to every patient.**



# Who Am I?

By Jennifer M. Loglisci  
West Springfield, Massachusetts



Jennifer and her sister Crystal.

**I am a young woman who has cystinosis.**

**Cystinosis does not have me.**

**I have always used cystinosis to drive me forward.**

**Growing up, it was a question of how long this baby girl was going to live?**

**Now it is about living the best life I can while reflecting on my experiences.**

**A**t 28, I have had many medical ups and downs, but I have remained optimistic about my future. Being diagnosed at 18 months old, my future was very gray, but my thirst for life was strong. I have battled through two kidney transplants, gracefully donated by my father and my cousin. Medication is a large part of my life and the side effects of some of these medications (such as Prednisone), have caused my bones to become soft and I have had multiple bone surgeries to help strengthen them. I don't want to dwell on negative aspects of living with cystinosis. I want to focus on overcoming the obstacles that cystinosis brings.

**"This baby isn't going to make it to her third birthday."** That's what my parents continuously heard from the doctors. As time went on, and the doctor appointments passed, doctors still said things such as, "This little girl isn't going to make it to the age of six." Every year the doctors were more stunned. Living with cystinosis is a daily struggle, but I believe that I have proved that I am a fighter, and always have been a fighter. Fortunately, the strength of my family and friends have given me strength.

I would not be who I am today without my family. So it is not hard to tell you about my inspiration. I can not imagine my life without my older sister Crystal. She is my role model and the wings that keep me moving. Crystal has been with me through thick and thin – by my bedside at home and in the hospital. You are probably thinking, "That's what a sister is supposed to do," but Crystal has always gone beyond normal sister limits. We fought like sisters do when we were young, but as we have grown older we have become more like best friends.

I think relationships between siblings when one is chronically ill are different than relationships in a healthy family. Maybe it is because we understand how important it is to work to achieve a good quality of life. Maybe it is that our family is more appreciative of our time together. I really don't know, but Crystal has taught me to fight for what I want in life. There are no words that truly define our relationship, but if I had to choose one, it would be "intertwined," because we are intricately linked to each other.

**My goals are always within reach because of my positive attitude.**

With my family's support, living with cystinosis has become nothing more than a routine. I get up every day, go to work, and attend college classes, all while taking my medication. As I have grown older, I realized the importance of taking my medications on time

and attending my doctor appointments.

I realize that I need to live each day to the fullest. Luckily, medication can travel. I have flown a plane, been skydiving, and traveled to Mexico, California and Brazil. I believe that it is the difficult days that make the normal, typical, boring days a little more beautiful. My goals are always within reach because of my positive attitude.



# with ~~Run for~~ Mary Head

*By Melissa Head, Mary's mom  
Yelm, Washington*

**During the first couple years after Mary was diagnosed with cystinosis many people told us that things would “get easier.” I’ve been thinking about that lately. Has it become easier? Or have we just become numb to it?**

**D**uring dinner recently, Mary threw up on her plate. This is by no means a rare thing, but it doesn't phase our family anymore. No one flinched or even stopped eating – except for me, to quickly replace her plate with a clean one with new food.

Mary knows when she is going to throw up so she just runs to the bathroom and gets her step stool ready. If the kids are watching a show together and Mary runs to the bathroom, they just pause it until she gets back. She just smiles and resumes her activity.

When we drive anywhere, Mary has a plastic cup in her car seat and I have a backup in the front for occasions when she gets sick in the car.

So yes, we've all become a bit numb to the ill effects of cystinosis. And to be honest, I am thankful for it.



On Father's Day, Mary made J.R. a card and she wanted me to write that the reason she loved her daddy was because "he was the best daddy in the whole world and he always gets her milk and water."

Mary can't wait to grow up and be as tall as me and as old as her brother and sister! This fall she had celebrated her fifth birthday and is starting preschool.



*Mary, front row, with friends Dawn Olson, holding baby girl Charlie, Aubree Olson, Kendall Olson, Reece Olson and Kevin Taft, who won the race.*

We homeschool, so we have all her preschool books in her special bin. She can't wait to learn! She wants to be a doctor and has a first aid kit that we replenish frequently with band aids because she is always "fixing us and making us feel better!" Mary also wants to be a butterfly though so...we'll see!

Mary is the perfect little sister – equally deciding each day if she is going to be on Audrey's side or Wylie's!

She makes us laugh – especially at dinner! She uses dinnertime to entertain us. She rarely eats – her choice for dinner is often milk and water so she uses the dinner table as a time of giggling and jokes!

McDonalds chicken nuggets are Mary's current food of choice! Her other staples are Doritos, cheese, vanilla yogurt, hot dogs, pizza and butter, which she eats by the spoonful. She stated the other day that "apples

are foul!" She has never eaten a piece of fruit.

Some days Mary eats and some days she doesn't. When she does eat, it isn't very much so thank goodness for her G-tube. She gets her daily nutritional and caloric needs and medications through it. Every

three hours throughout the day Mary takes vitamins and every waking hour she receives eyedrops. It's quite a regimen for a little gal, but so far she takes it all in stride.

It's been an incredible year for raising money for cystinosis. Last year, Music for Mary was successful and fun, and we are currently working on round two!

One day during Mary's gymnastics class, her teacher, Kelsey Nore, approached me with an idea she had for a fundraiser at her high school for cystinosis.

*Continued on next page*



## Run for Mary Head *Continued from previous page*



*Best Friends Forever: Brynn Burt, Ashley Rager, Mary and Londyn Walker.*

I was invited to speak to her class and explain the disease. Mary bravely, went in front of the class and showed them how she gives vitamins to her baby doll with a syringe. Her baby doll has a tubie like Mary's! The class was receptive, compassionate and excited about fundraising for cystinosis.

These teens wanted to do something big and were eager to help in every way they could.

Many in the class spent well over an hour visiting with me and playing with Mary.

Plans were underway! In April, Kelsey and her leadership class put on a benefit concert using local singing talent from the school, all you can eat spaghetti feed, auction and raffle. Kelsey and her class put their heart into the evening and it was awesome! Mary has made a lifelong friend in Kelsey!

**“What is the name of that disease I had when I was a little girl?”**

Run for Mary – a 5k race was our second fundraiser this year. Al and Julie Best, our friends from church, approached us about putting on a race in our town of Yelm. They spent hours getting local sponsorships, road crews and permits organized and so much more! The race was

a big hit, with about 55 runners. Most everyone stayed after to hear the results of the top female and

male finishers and also to see who won the raffle prizes that were donated by local businesses. We are so grateful to the Best family for all the hard work and time they donated to help raise money for cystinosis research.

As I write this, J.R. and I are anxiously waiting

to hear if our insurance company has approved Procysbi™ for Mary. We know it will change the quality of her life drastically, and it can't come soon enough. We are a family that lives day to day, and we have faith and hope for a cure. The days that are hardest on Mary, are the days when our hearts are heaviest. We trust in the Lord for comfort and strength.

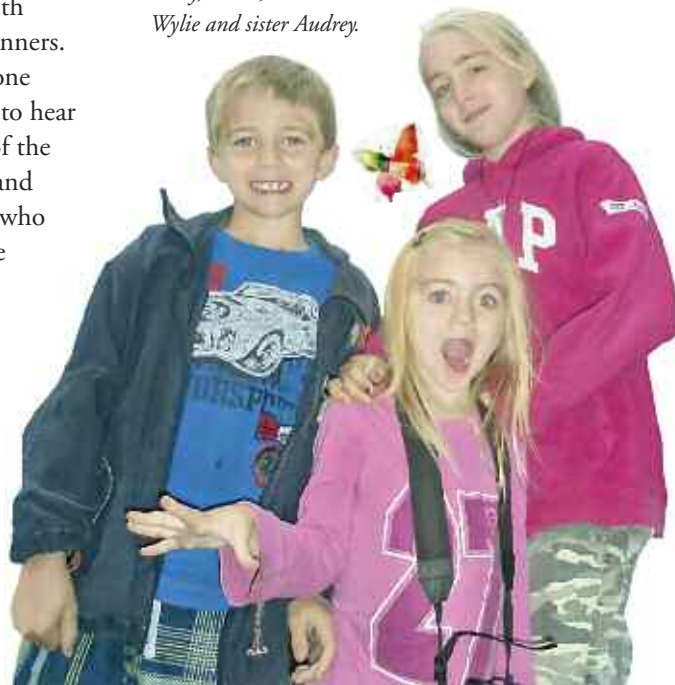
We are so incredibly thankful for the Stack family and everyone involved with the CRF including the researchers who are working tirelessly to find a cure. They are all in our daily thoughts and prayers.

I will never forget calling the CRF after Mary was diagnosed and leaving a rather desperate message. Nancy called me back so quickly and was a great comfort to me in every way!

Mary is a such a happy child, finding joy in almost everything. She is rarely without a smile. She is smart and witty and outspoken. She is a light and joy in all of our lives.

As another mother once stated, I hope some day Mary can approach us and ask, “What is the name of that disease I had when I was a little girl?”

*Mary, center, with her brother Wylie and sister Audrey.*





# Joshua Clarke: The Happy Trooper

**Joshua, our youngest child, was diagnosed with cystinosis in January 2008. It doesn't seem possible that it has been five and a half years since this horrible disease abruptly changed the course of our lives. The stark contrast between then and now is equally as unbelievable.**

In the months that followed Joshua's diagnosis, we were without hope and knew only despair. We lived in fear of the unknown, constantly contemplating what devastating effects this disease was inflicting on our young son's body. It is amazing to look back at pictures of him at 15 months of age before his diagnosis and to look at him now. That hopelessness and despair has turned into hope and joy.

Joshua is now a thriving six-year-old, doing extremely well and keeping himself quite busy. He remains a loving, spirited and joy-filled little boy. His positive energy and determination keeps our family strong with lots of smiles and laughter. Joshua keeps himself busy with lots of different activities. This year he played basketball and soccer for the first time and continued in baseball. He was asked to be part of the All Star team in his division for baseball and is known for his lack of fear on his soccer team. Generally you will find him defending the largest boy on the opposing team during any given Saturday morning soccer match. Joshua also enjoys reading, Cub Scouts and quality time with his brother and sister. He has been blessed with two wonderful siblings who want to play with him always and take excellent care of him.

Currently, Joshua takes eight different medications a day, in addition to a nightly growth hormone injection. This summer Joshua was very blessed to start on the recently FDA-approved, sustained-release medication Procysbi™. This vital medication is taken every 12 hours, and replaced Cystagon®, which was taken every six hours. We now manage all of his medications during the day and he receives no medication throughout the night. This is an incredible improvement to our family's quality-of-life ...D.J. and I can now sleep through the night! In addition, this summer



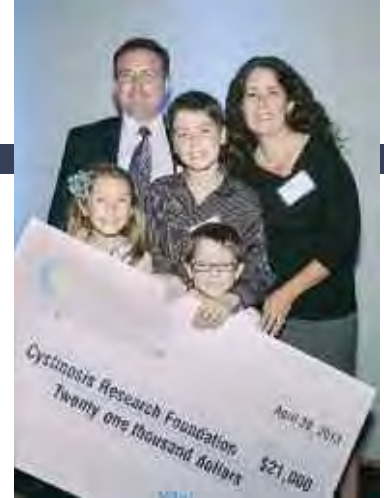
Joshua was accepted into the Children's Hospital of Orange County Multi-Disciplinary Feeding program which specializes in weaning children off their G-tubes. The program requires a 19-day inpatient stay where the focus is solely on overcoming feeding difficulties through a variety of disciplines. Joshua did amazingly well and is now able to get most of his calories through oral feeding.

Although Joshua is doing remarkable and his quality-of-life has greatly improved, cystinosis has a way of keeping reality in check. As I watch Joshua during his various activities, I cannot help but notice that he has to work twice as hard as other children to kick the ball hard, run fast or write a sentence. I am constantly reminded that cystinosis affects him each and every day. We place our confidence in the life-saving medication, Procysbi™, which we hope will continue to prolong his life and prevent the horrible effects of this disease from causing Joshua any further damage.

We are so thankful to our donors and to the Cystinosis Research Foundation for all their generosity and support.

It is through the efforts of many that incredible strides have been made for these brave patients. We are confident that continued improvements will be made toward a better quality of life and a cure for all cystinosis patients will be achieved.

Thank you for your support of our family, Joshua's Journey of Hope and the Cystinosis Research Foundation.



*The Clarke family presenting a check to CRF at the 2013 Natalie's Wish event.*

Photo by Bob Hodson

*Joshua Clarke really is a happy trooper.*

NOW AVAILABLE!

# CYSTARAN™

(cysteamine ophthalmic solution) 0.44%

**S**igma-Tau Pharmaceuticals, Inc. (Sigma-Tau) announced the commercial availability of CYSTARAN™ for the treatment of corneal crystals in patients with cystinosis on May 1, 2013.

Corneal cystine crystal accumulation is a common symptom of each of the three forms of cystinosis – nephropathic, intermediate and nonnephropathic.

The formation of cystine crystals within the corneas of the eyes may lead to photophobia, or sensitivity to light. Other effects include eye pain, damage to the corneas, foreign body sensation (the feeling of something in the eye), and squinting. These may be debilitating to the patients who are affected by the crystals.

CYSTARAN™ is the first and only FDA-approved therapy for the treatment of corneal cystine crystal accumulation and is available to cystinosis patients in the U.S. with a valid prescription.

The most frequently reported ocular adverse reactions occurring in ≥10% of patients were sensitivity to light, redness, eye pain/irritation, headache, and visual field defects.

Patients, caregivers and physicians in the U.S. can call the CYSTARAN™ Hotline (1-800-440-0473) to obtain information about ordering CYSTARAN™ through Accredo Specialty Pharmacy. The hotline is staffed from 8 am–5 pm Central time with customer care representatives, pharmacists and nurses to process, counsel and answer any questions you may have about CYSTARAN™.

Once an enrollment form is received at Accredo, representatives will research benefits for CYSTARAN™, explain any financial responsibility patients may have and directly refer patients and caregivers to assistance programs available for copay assistance and assistance programs for those patients who are uninsured or underinsured to determine eligibility.

Upon the fulfillment of the prescription, the delivery of CYSTARAN™ will be direct to the patient. Due to the temperature sensitivity of CYSTARAN™, it is shipped in a custom shipping container allowing for dry ice to keep the CYSTARAN™ frozen for up to three (3) days. In the box will be a special temperature monitor that will be able to tell you if the product went out of the defined temperature range during the shipping process. This allows the product to be shipped all over the U.S. and stay within the specified storage temperatures.

Once delivered, CYSTARAN™ bottles should be stored, unopened, in the freezer. When ready for use, thaw 1 bottle for approximately 24 hours (do not heat the bottle or its contents). Thawed CYSTARAN™ can be stored between 36°–77° F for up to one week. Once a bottle has been opened the CYSTARAN™ solution is only stable for 1 week and the bottle should be thrown away after being open for 7 days no matter how much medication is used. There is a place on the bottle to write the discard date as a reminder.

CYSTARAN™ should be given as 1 drop in each eye every waking hour, unless your doctor gives you other directions. If you wear contact lenses, you should remove your contacts before using CYSTARAN™. You can reinsert your contact lenses 15 minutes after administration of CYSTARAN™.

You will receive a reminder call from a specialist at Accredo when you are due for a refill, but are always welcome to call the hotline with any questions.

**Sigma-Tau remains dedicated to cystinosis patients, healthcare providers and the community through continued communication, education, support and access.**



To obtain more information about CYSTARAN™ please call Accredo Specialty Pharmacy: 1-800-440-0473. To contact Sigma-Tau, please contact Lesli King, Senior Manager, Patient Affairs: 1-301-670-5450 or Lesli.King@sigmatau.com. Please see full prescribing information at [www.cystaran.com](http://www.cystaran.com)

 **sigma-tau**  
PHARMACEUTICALS, INC.  
**Rare dedication**

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



Chloe and Gabbie with their mom Jody Strauss.

Photo courtesy of Mark Essner



Plates  
&  
Dishes

Not  
Medications  
&  
Supplements

# Gabbie Strauss

Gabbie is now a grade 1 homeschool student who is flourishing in her new learning environment. She is joyful, energetic and determined. She loves to be outside, she loves to swim, she loves ice cream and she really loves her younger sister Chloe and new baby cousin, Jacob.

Gabbie's smile makes me smile, her laugh makes me laugh. She is an amazing gift and we feel so blessed to have a daughter like her. When cystinosis tries to bring us down, we fight back with faith. When I look at our medicine cupboard (that's right, a full cupboard!) I thank God that one day there will be plates and dishes there instead of countless bottles of medications and supplements.

When we find her bed sheets and pajamas wet, I thank God that it won't be like this forever, it will end. Someday soon, the sheets will be dry, there will be no more diapers, no more poise pads, no more medications and no more cystinosis.

I am so grateful and thankful for the Cystinosis Research Foundation and everyone who supports cystinosis research. Keep believing and praying for a cure!



# It Takes a Village

Four years ago our family started Cystinosis Awareness and Research Effort. Our goal was simple, to raise money for cystinosis research. Our first year of fundraising included over six different fundraising events. This tradition has continued. We'll do anything and everything to raise any amount for cystinosis research. We are so thankful for amazing family and friends who faithfully support Gabbie and cystinosis research. We couldn't do this on our own. Here is a breakdown of our fundraising effort over the last year.



## Baby Shower

Baby showers are supposed to be all about the mother-to-be and baby on the way. As a new mom, you need lots of items to support your little one. In November 2012, Gabbie's Aunt Jill decided to put Gabbie's needs ahead of her own and turned her baby shower into a cystinosis fundraiser. The invitation read:

*We are pleased to invite you to this baby shower. Please do not bring baby gifts or a flower. Instead please bring a monetary gift, a donation please, to give to our sweet niece's rare disease.*

Aunt Jill's love helped raise \$975 for cystinosis research.

## UNCLE JOHN'S PIG 'N POKER

Gabbie's 'Uncle John' loves poker, pig roasts and Gabbie.

Although it was a cold day in April for an outdoor pig roast, hearts were warm. Friends of John and Robyn Beresford braved the weather and gladly contributed an amazing total of \$975 for cystinosis research.



## Lance Roberts Shredding Party

Even the boring task of managing unwanted documents can be an opportunity to raise money.

Our family spent an entire day in June with friend and realtor, Lance Roberts, and Integra Document Shredding. Lance welcomed clients and friends with coffee, treats and a free drawing, while they brought unwanted documents and a donation to cystinosis research. The event raised more than \$600.





Gloria Deutschlander, Carol Stacey and Dianne Strauss teamed up again to plan and execute another fun filled evening of fashion and food with all proceeds going to cystinosis research. In May, almost 300 women attended the show and enjoyed fashions by Taya and Seasons Fine Clothes, appetizers by Chef D, decadent cupcakes by Marianne Peters of Sweet Mary's and a diverse silent auction table that included more than 75 items. Everything for the event including the venue, food, entertainment and gift bags were all donated. It was an evening enjoyed by all and it raised \$24,145.



*Fashion show organizers Carol Stacey, Gloria Deutschlander and Dianne Strauss*

## GRANDMA'S Summer Sausage Fundraiser



Gabbie's Grandma, Dianne Strauss, has made finding a cure for cystinosis a priority. Dianne works hard all through the year organizing, planning, obtaining food and silent auction items to make every event successful. But that's not all! In the last year, Dianne has sold 275 sticks of summer sausage and made \$922 for cystinosis research. What an amazing grandma!

## Street Impressions Charity Car Show and BBQ

The 4th annual Charity Car Show and barbecue was a two-day event in July that attracted over 100 cars and many spectators. Gabbie's grandma, Dianne Strauss, had all of the food and supplies for the barbecue donated, so every dollar made went to cystinosis research.





*Jenna and Patrick at a Tree Top Adventure Park, Lake Tahoe, California, August 2013.*

# SOAR HIGH TOWARD A CURE

## JENNA AND PATRICK PARTINGTON

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



Jenna and Patrick during the final trial visit with nurse Cindy Steffel.



### Dear Supporters of Jenna & Patrick's Foundation of Hope,

We are once again receiving Jenna's and Patrick's lifesaving cystinosis drug by mail! Last month we said a tearful goodbye to Dr. Paul Grimm and the kids' much-adored nurse, Cindy Steffel at the Clinical Trial and Research Unit at Stanford University.

We are so grateful to the Cystinosis Research Foundation for their commitment to funding all of the research that led to this slow-release drug formulation, which vastly improves the quality-of-life for cystinosis patients and their families. We are also grateful to Raptor Pharmaceuticals for bringing Procsybi™ through FDA approval and to market, and for continued efforts to ensure that all cystinosis patients can enjoy the drug's benefits.

Jenna and Patrick will be nine years old on December 7. Their eighth year was one we truly enjoyed as parents, as we watched them become more independent, making every area of life just a little bit easier. We marvel at them as they learn to read and write, and become little people!

Patrick has given up liquid medication altogether, and Jenna is down to only Cytra-K in liquid form. They swallow 19 pills each night and 13 in the morning... and are at times known to walk to the medicine cupboard, open the bottles and count and take them by themselves! It's a far cry from the days they when we would lay them down, restrain their arms and legs, and use a dropper and a lot of tough love to get their meds down each of their throats twice a day.



### Breakfast of Champions? You Bet!



*Jenna and Patrick's daily medicine routine has certainly gotten better but it's still far from easy. And it's not one many of us could easily tolerate. Patrick takes all pills, Jenna still takes liquid...served up in espresso service!*

# SUMMER FUN WITH FAMILY AND FRIENDS



*Hadley Alexander, who was diagnosed with cystinosis on April 4, 2012, relaxing with her sister Stella, and Jenna and Patrick in McCall, Idaho this summer.*

**This summer, we had fun cooking, golfing, swimming, canoeing and biking as we spent time at home and traveled to Idaho and Lake Tahoe. (When you have two kids with kidney disease, road trips are a never-ending adventure in “jungle potty!”)**

We spent a special summer weekend in McCall, Idaho, with Ben and Marcu Alexander, and their daughters Stella and Hadley. We met the Alexanders one year ago at the CRF Day of Hope Conference, after their youngest daughter, Hadley was diagnosed. Cystinosis is very rare and very scary, and we find it comforting and enlightening to spend time talking with and enjoying the company of others who are dealing with it, too.

As the kids grow and change, so too does the way cystinosis presents in their bodies. Since their diagnosis at 15 months of age, Patrick and Jenna have been followed by a pediatric nephrologist (a kidney doctor), and a pediatric ophthalmologist. The first parts of the body

to be affected by cystinosis are the kidneys and eyes, and much can be determined about the progression of the disease and the effectiveness of treatment by having doctors keep a close watch on them. However, cystinosis is a progressive condition that wreaks havoc on every system of the body, and while Procysbi™ works to keep the disease at bay, it is by no means a cure.

This year saw us visiting new doctors in new specialties who will tend to the ever-evolving state of Jenna and Patrick’s health. We visited Shriners Children’s Hospital for orthopedics (knock-knees and flat feet), a dietician



*With their big cousins Palmer Batt (L) and Spencer Batt, during summer break in McCall, Idaho. Palmer works for Microsoft and makes regular donations to JPFH, which are matched by his company! Last year he made a \$1,000 donation after returning from his third tour of duty: two in Iraq, one in Afghanistan.*

(bizarre eating habits), a pedodontist (adult teeth are coming in with no enamel), and the kids will soon begin seeing an occupational therapist to help deal with challenges at school. I spend time overwhelmed by these new challenges that present themselves, but Kevin reminds me they are all “fixable” things. There is a minor surgery to straighten knees. The dietician has already proven helpful. Unsightly teeth can be bonded. And while schoolwork is a challenge, the kids love their friends – and are loved by their friends – and they enjoy their teachers and school environment immensely. Jenna and Patrick are happy and sociable. We’ll take it!

*Jenna with her classmate and friend Ana.*







# KICKIN' UP DUST FOR A CURE

Jenna with country music singer/songwriter Jon Pardi.

Brett Eldredge, on stage at the Golf & Guitars event, is a 2013 nominee for CMA (Country Music Association) Best New Artist.



Ron Thomas, Jenna and Patrick's godfather, is one of the founders of JPFH. Ron is also Kevin's business partner of 18 years, neighbor and dear friend.

## Jenna & Patrick's Foundation of Hope had a great year of fundraising.

As fans of country music, our family loved being part of Sacramento's Annual Golf & Guitars event last May. With the help of CBS Radio, Haggin Oaks Golf Course and the Morton Golf Foundation, JPFH raised \$42,000, which will be passed on to the CRF for research.

Thank you to our dear friends, Byron and Gabrielle Kennedy and our new friends at Morton Golf Foundation for making us a part of this event.



Teresa with country singer Brett Eldredge (L) and Byron Kennedy. Byron works for CBS Radio and helped the foundation land the golf & guitars gig.



## Party Hearty for a Cure

Planning is already underway for next year's *Swing & Bling* fundraiser, to benefit Jenna & Patrick's Foundation of Hope. Golfers will enjoy the *Swing* portion of the weekend at Teal Bend Golf Course on Friday, February 21. The *Bling* dinner event will once again be held at Sacramento's beautiful Citizen Hotel on Saturday evening, February 22.

Visit [www.jennaandpatrick.org](http://www.jennaandpatrick.org) for details.



# An Ongoing and Positive Journey OF ACCEPTANCE TO LIVING WITH CYSTINOSIS

Photos by Lars Wänberg



## CELEBRATING EASTER WITH SPECIAL FRIENDS

This spring, our family made the three-hour drive to Clarkston, Washington to spend Easter with the Flerchingers. We have always been close to the Flerchingers. They were the first people we met living with cystinosis. Tina was about five and Henry about two when we met around Christmas in 2008. The Flerchingers have taught us so much and are so generous about sharing information and supplies they no longer need. We love them and the pictures of Tina and Henry from over the years are precious.



## A LONG FUN-FILLED SUMMER

We had a great summer. The weather was hot and our summer started early and lasted into September. We don't have long summers in the Northwest, so we are always grateful when we get a long one. We did a lot of boating and swimming, and we made our annual weeklong family camping trip to Priest Lake in northern Idaho.



## RACING FOR AWARENESS

Race Across America, also known as RAAM, has been described as the greatest bicycle race in the United States! Over 30 years, RAAM has become an international attraction for cyclists and a leading platform for raising money for charitable causes.

An incredible team of cyclists from our hometown of Sandpoint, Idaho, rode RAAM in honor of 24 Hours for Hank, and to raise money and awareness for cystinosis. For 7 days, 1 hour and 42 minutes, Jacob Styer, Melvin Dick, Dean Kyriakos and Alan Lemire rode non-stop across the country wearing their cystinosis jerseys. We are grateful for their efforts and commitment to finding a cure.

RAAM is more than a bike race. It is about ordinary people being great.



Left to right: Melvin Dick, Alan Lemire, Dean Kyriakos, Hank Sturgis, Jacob Styer.

It's about realizing dreams, a journey of the heart and soul. It's about being the toughest of the tough. It's about camaraderie, teamwork, and the incredible RAAM family. It's about taking the stage for a charity or cause that matters, to raise awareness and funds. It's about the vastness, richness, beauty, and generosity of the United States and the people who live here. It's an experience never to be forgotten.

Race Across America endures due to its amazing effect on the human consciousness and for its incredible

feats of will power, inspiration, and heart. The race starts in Oceanside, California and ends in Annapolis, Maryland. The ride is 3,000 miles, touching 14 states and climbing over 100,000 feet. Teams typically cross the country in 6 to 9 days, averaging 350 to over 500 miles per day. Solo racers finish in 9 to 12 days, averaging 250 to 350 miles per day. Teams have a relay format and race 24 hours a day. Solo racers have the challenge of balancing a few hours of sleep each night against race deadlines.

## READING, 'RITING AND TRYING REALLY HARD

Henry started first grade this fall at Farmin Stidwell Elementary. He is doing well academically but requires a lot of extra help in certain areas. We are very proud of him for reading his practice books and he has mastered many of his sight words. He still receives Occupational and Physical therapy during school, a half-hour each week. He loves PE and playing a computer learning game his teacher Mrs. Dash developed.

In an effort to help with any odors associated with cystinosis, our school professionals came up with two great ideas: they let him chew gum and have smelly lotion at his table. It is still a challenge to keep Henry healthy from colds and germs so he does not miss school. There is also the "playing hooky" we encounter when he feels sick at school and wants to come home. We have to determine whether he is really sick or just trying to get out of a hard day.

Henry has anxiety at school, which is hard work for him. He so desperately wants to keep up with the other kids. But how Henry feels on a typical school day compared with healthy kids can be very different.



## EXIT FROM STANFORD CLINICAL TRIAL FOR RP-103

On October 4, we made our final trip to Stanford for our exit exam on the RP-103 clinical trial. We are now using Procybsi™. We are grateful to have been on the study and for the clinic team including Dr. Paul Grimm, Suvarna, Allison, and Cindy, our favorite nurse for blood draws.

We always looked forward to seeing the families we already knew, as well as meeting new ones. Henry loved helping his buddy Sam if he was in clinic with us. He would help the nurse take Sam's temperature, and help in any way he could. Henry was extra brave with his blood draw to help Sam with his.

Dr. Grimm will soon be starting a cystinosis clinic at Stanford. Anyone is welcome to visit; cost and other details will be available soon. The clinic will be especially valuable for patients who live in remote areas where a nephrologist is unavailable.



Photo by Lars Wänberg

## FAMILY MATTERS

### CONNECTING

Relationships with the ones you love provide so much support. Yet, it isn't always easy. Balancing all of the demands of life in general – managing prescriptions and doctor's appointments, while making time for your spouse and close friends – can be hard for parents.

We constantly remind ourselves to provide each other love, friendship and motivation. Sometimes the little things mean the most, and often keep your relationship strong.

### PREPARING FOR THE FUTURE

Tyler Carlsen, a chronically ill teen, offers sage advise when he writes:

**“We have the ability to begin and maintain a completely normal and happy life no matter what the circumstances may be, we just have to be prepared to face any challenges that we may encounter and stand strong, knowing that those challenges are only temporary.”**

Henry recently had a bad infection with a temperature of 103.5 and had to go to the Emergency Room in the middle of the night. I took a picture of him and his Dad sleeping in the hospital bed. These things happen from time to time, and I guess this is an example of a challenge that is only temporary. You just pick up and move past it.

*Sincerely written by Tricia and Brian Sturgis*



# Cowboy Up



# for a Cure

## 24 HOURS OF SCHWEITZER – MARCH 21 AND 22, 2014

Plans are well under way for our next 24 Hours of Schweitzer. In 2014, we're adding games and scavenger hunts to keep skiers going through the entire 24 hours. It is usually cold in the middle of the night and any ideas we come up with to keep the event entertaining enhances the skier's experience.

Skiers of all ages participate. Some are families – parents with their kids, and others ski solo. Some are very competitive teams, and some just ski for fun. We have participants and sponsors from all over the United States, and donors who pledge skiers in support of their efforts from around the world, including Australia, Mexico, England and Canada.

The ski event is followed by an amazing awards dinner and auction on Saturday night. Many people from our community, some who may never ski, come to celebrate and enjoy the evening. They are eager to learn about cystinosis, what Henry's life is like, and what our family does to care for him.



***We hope you will join friends old and new from around the United States in March 2014 as we Cowboy Up for A Cure.***

***Visit [www.24hoursforhank.org/events.html](http://www.24hoursforhank.org/events.html) for details.***



*Title sponsor*





# LIBRARY SCIENCE

## Dr. Francesco Emma combs through a warehouse of molecules, searching for better cystinosis treatments.

There's more than one way to attack a problem, including an orphan metabolic disease like cystinosis. No one knows this better than Dr. Francesco Emma, a researcher and physician who is dedicated to turning discovery into better cystinosis treatments.

"One approach is to take a model of a disease and try a drug that you think will work," said Dr. Emma, head of pediatric nephrology at Bambino Gesù Children's Hospital in Rome, Italy. "Another approach is to try *every* drug."

Thanks to a grant from the Cystinosis Research Foundation (CRF), Dr. Emma is searching through drug libraries for a multitude of options in the fight against cystinosis. Some of those libraries contain as many as 20,000 molecules, offering the biggest opportunity possible to test against models of disease.

But Dr. Emma has chosen to focus on a smaller library, which contains 1,280 small molecules.

"This specific library is interesting because it only contains molecules already licensed for human use," the doctor said. "With a rare disease like cystinosis, that's very important, because it's incredibly expensive to develop a new drug."

Dr. Emma's research has identified 20 molecules that he calls "very interesting" as potential cystinosis treatments. These molecules have been shown to decrease the cystine content in cells by 60 to 70 percent, "and now we need a second round (of testing) to see if we can repeat these results," he said.

The doctor hopes that eventually one or two drugs will stand out as especially effective, and they'll advance to testing using the mouse model that has been so critical in the research battle against cystinosis.

The current step is kinetic testing to determine effective concentrations of drugs and frequency of administration. Even if such testing doesn't reveal a breakthrough treatment, it might identify one that can combine with cysteamine to enhance current protocols.

"We hope that by early next year the kinetic studies will be completed, and in the next two years animal studies will be performed," the doctor said.

Multiple tracks of cystinosis inquiry have been at the center of Dr. Emma's professional life for a decade.

"I've always been interested in tubular disorders, and cystinosis is clearly one of those," he said. "It's particularly interesting for me because cystinosis is still not well understood; we're still looking for a thorough understanding of the disease."

The support of the CRF and the cystinosis community also fuels Dr. Emma's focus. "The CRF is instrumental because everyone is organized and dedicated,"

he said. "It's only through the CRF's capacity for raising funds and sponsoring research that we've been able to see such great progress."

The advances Dr. Emma has seen include the discovery of the cystinosis gene, the development of the mouse model by Dr. Stéphanie Cherqui, University of California, San Diego, and the promise of delayed-release cysteamine.

"There's also incredible hope for bone marrow transplant," he noted. "And then there's just the overall awareness of the disease. We're diagnosing earlier than ever, and that's critical to effective treatment."

As he both researches and treats cystinosis, Dr. Emma takes on the added role of evaluator, serving on the CRF Scientific Review Board. He and his international colleagues research proposals for their potential to improve lives. In so doing, he also improves his own global perspective.

"This role allows us to increase our contact with experts in the field," he said, "and such collaboration is very important to our research."

The future of that research is bright, he added – the potential for new drugs and new understanding a beacon on the horizon.

As close as the local library.



# CCIR LOOKS TO THE HORIZON FOR ADVANCEMENTS IN CYSTINOSIS

**Thanks to recent scientific advances, milestones in the treatment of cystinosis that once seemed far away now appear to be within reach.**

**As covered in this issue of *Cystinosis Magazine*, incredible improvements in cystinosis care and therapy have been achieved with the advent of delayed-release cysteamine.**

**A**lso highlighted in this issue are the sophisticated scientific and technological applications in cystinosis research, such as gene therapy and nanowafer drug delivery to the eye, that are on the horizon. The goal of CCIR is to support these efforts and hasten progress towards a cure.

CCIR collects accurate and current information about the disease from cystinosis patients and their families. The idea is to have this information available to present to important policy makers, funding institutions and regulatory agencies such as the Food and Drug Administration (FDA) who play a role in determining the future of cystinosis research projects. Ultimately, the cystinosis community itself can provide compelling evidence for continued funding and investment of resources in research for this rare disease. Therefore, the active and continued participation of all individuals affected by cystinosis around the world is desirable.

## WHERE ARE WE NOW?

CCIR is now 415 people strong with representation in 37 countries. The professionals who request access to this anonymous database are as diverse as the patients represented therein. The 68 registered professionals range from treating physicians, nurses, scientists and pharmaceutical representatives, and they come from 20 countries around the globe.

## WHAT TO EXPECT NEXT FROM CCIR

We are thrilled to introduce a Dutch translation of the CCIR website. CCIR is now available in five languages including Spanish, French, Portuguese, English and Dutch. Thanks to the tireless efforts of a few people in the Netherlands who volunteered to translate the entire website, registrants in the U.S. and elsewhere can now compare their profiles with Dutch-speaking registrants.

There will soon be a revised CCIR survey. This summer, experts from around the world were asked to contribute relevant questions that would improve our understanding of

the disease. The revision is necessary to reflect the changes in treatment options and to allow the capture of more detailed information about the medical history of cystinosis patients. Please be on the lookout for a notice of publication and complete the supplementary questions as soon as you have a chance, or when its time for your next yearly update.

## THINGS TO REMEMBER

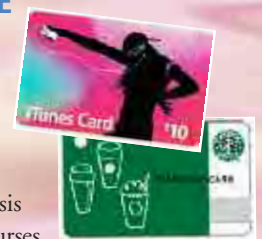
Aside from the knowledge that you are a part of a worldwide effort to find a cure for cystinosis, registration brings added benefits. Don't forget our *Ask an Expert* feature that allows you to anonymously submit questions about cystinosis to medical experts. See the next page for a few previously answered questions.

If you are considering participating in a clinical trial, resources are available on the website to help you make informed decisions. Announcements and links to upcoming clinical trials are also listed for your convenience.

## PARTICIPATE, UPDATE & ANTICIPATE

In the last issue of *Cystinosis Magazine*, we introduced the CCIR mantra: **Participate, Update & Anticipate the Next Medical Breakthrough**. This mantra should serve as a reminder of why your initial and continued participation in CCIR is crucial to making progress in cystinosis research. Spread the mantra to your physicians, nurses and other families you know who are affected by cystinosis so that the pace of registering and updating stays strong.

We would also remind you that *CCIR's Participate and Update Gift Card Campaign* is still in effect. We have \$10 gift cards to Starbucks and iTunes that we will send to all those who newly register or update their CCIR profile.



**CURE CYSTINOSIS  
INTERNATIONAL REGISTRY**

thank you!

We wish to send a special thanks to the CCIR Advisory Board members, made up of clinicians and patient representatives, who oversee the registry.

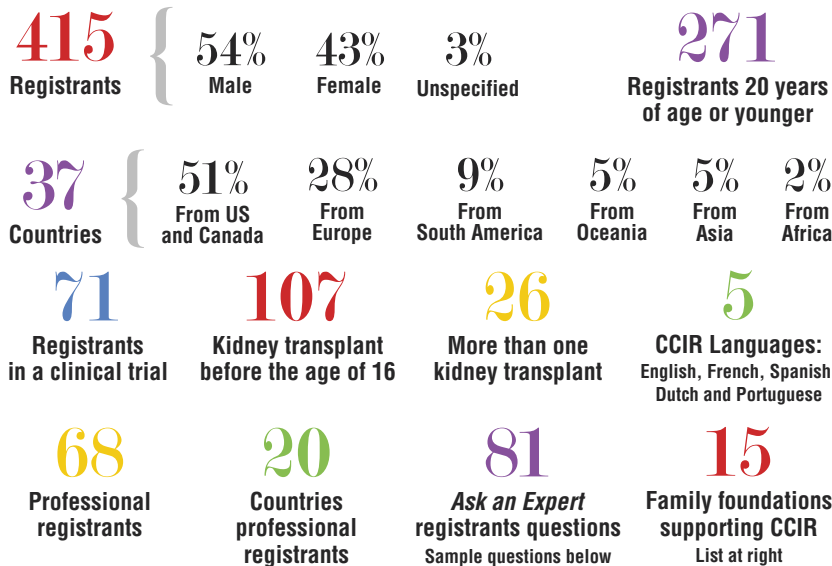
We would also like to recognize the volunteer translators who were integral in getting CCIR up and running in various languages: Dr. Vera Koch of Brazil, Dr. Edward Noordijk and Marjolein Bos of the Netherlands, and Dr. Reyhan El Kares of Canada and France.

And last but not least, we extend thanks to our wonderful participants without whom CCIR would not be possible!



## A World of Numbers

### QUANTIFYING CCIR'S SUCCESS



## Q&A

## ASK AN EXPERT

### Are there any dietary restrictions for children with cystinosis?

Many families worry about nutrition in cystinosis patients. A recommended approach to diet is not to restrict any type of food. Children with cystinosis have a difficult time getting enough calories and anything they eat is good. It is important to provide lots of salty food – some children grow better with a salt supplement.

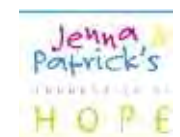
### Is it typical for young children with cystinosis to experience speech delays?

Early delays in some aspect of development are not uncommon in children with cystinosis. Speech may be one of those areas, since children with cystinosis may have problems early on with coordinating the muscles of their mouths and tongues. Speech therapy focusing on oral motor improvement should help. Most children with cystinosis will outgrow these early delays, especially with intervention.

### My child is on potassium citrate, but other children are on potassium chloride. What is the difference? Which do you recommend for cystinosis patients?

Potassium citrate can kill two birds with one stone. The potassium replaces potassium lost in the urine. The citrate is metabolized by the body, generating bicarbonate to neutralize acidosis – very useful for a child with cystinosis. However, if the proportions of these need to be adjusted separately, your physician may prescribe potassium chloride and some form of citrate or bicarbonate separately.

## CCIR PARTNERS AND ADVOCATES



Register at [www.cystinosisregistry.org](http://www.cystinosisregistry.org)



# Night of A Thousand Stars

**2014 NATALIE'S WISH CELEBRATION**

*The year's most important, most inspiring, and most fun-filled celebration*

**SAVE THE DATE**

*Saturday, April 5, 2014*

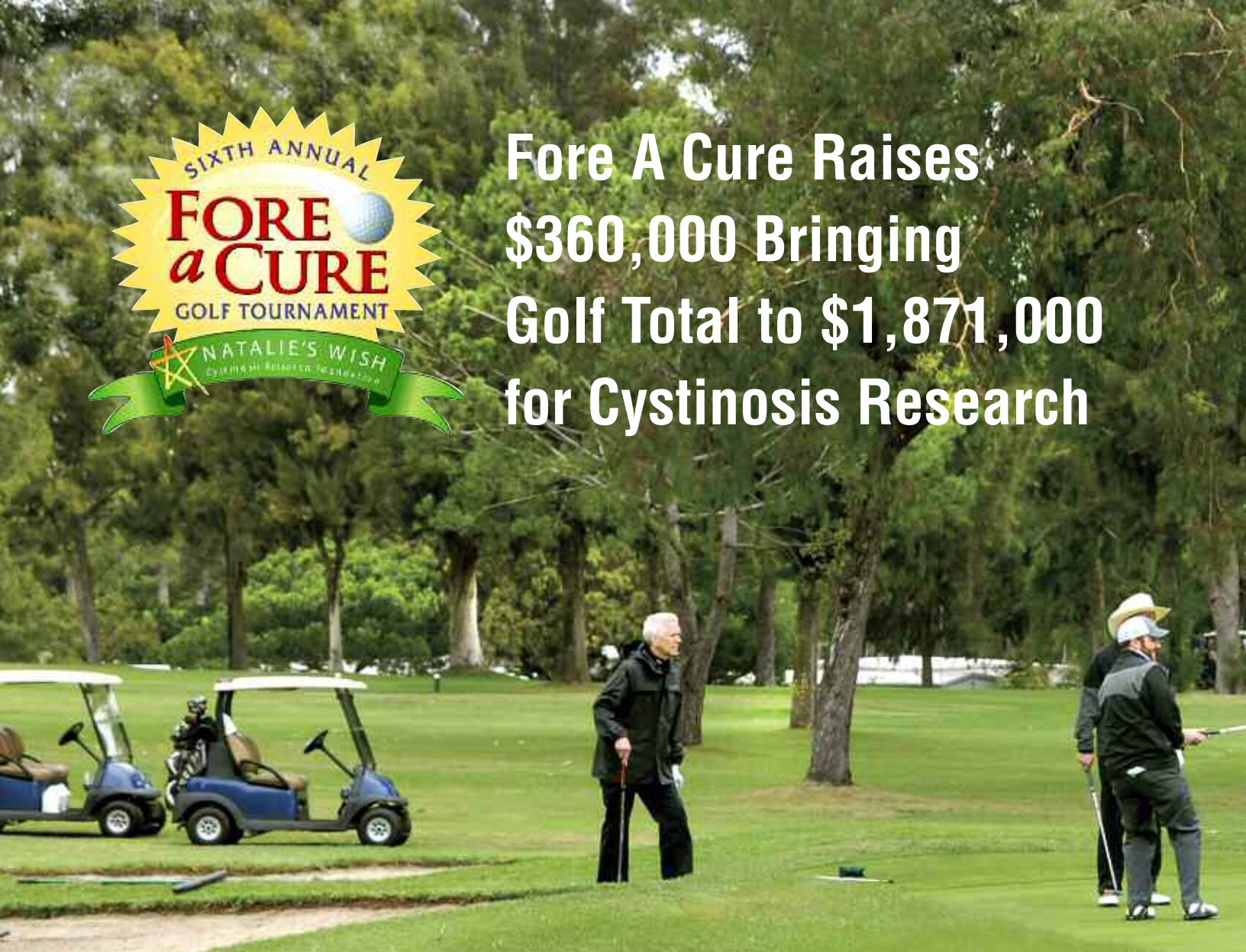
*Balboa Bay Resort, Newport Beach*



For sponsorship opportunities or tickets, contact Zoe Solsby  
at 949.223.7610 or [zsolsby@cystinosisresearch.org](mailto:zsolsby@cystinosisresearch.org)



# Fore A Cure Raises \$360,000 Bringing Golf Total to \$1,871,000 for Cystinosis Research



Mark your calendar for next year's tournament on  
Monday, October 27, 2014, at the Santa Ana Country Club.  
For information contact Zoe Solsby at 949.223.7610  
or [zsolsby@cystinosisresearch.org](mailto:zsolsby@cystinosisresearch.org).



Celebrating its sixth year, the Natalie's Wish Fore a Cure Golf Tournament had another record-breaking year at the exclusive Santa Ana Country Club. Led by its dynamic Chairman Vince Ciavarella, a powerhouse golf committee, a large group of dedicated volunteers and the generous sponsorship support of nearly 200 companies and individuals, the tournament raised \$360,000 for cystinosis research.

"The exceptional generosity and support of our friends and community has enabled CRF to make significant advances in treating cystinosis. CRF-funded researchers at institutions around the globe are making breakthroughs toward a cure. We are honored to be a part of the research progress," said Vince.

The tournament's reputation as "the best in Orange County" was confirmed as the event sold out three months prior to the Monday, October 28 shotgun. The threat of rain didn't discourage the 152 players from enjoying the fun-filled day of first-class golf, prizes and an opportunity to make a wish come true for those with cystinosis.

There were many past sponsors and golfers, as well as many new supporters and we are grateful to everyone who participated. Since 2007, the Fore a Cure golf tournaments have contributed more than \$1,871,000 to cystinosis research and the quest for a cure.

A dynamic committee and dedicated group of volunteers, most of who have been involved since the beginning, are the driving force behind the tournament. Their efforts made our 2013 event the best ever.





# *Congratulations and Thanks*

Nancy and Jeff Stack and the Cystinosis Research Foundation thank the sponsors, underwriters and participants of the 2013 Fore a Cure Tournament for making the event – which raised more than \$360,000 – our most successful ever.



## PRESENTING SPONSOR



## SILVER SPONSORS



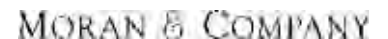
## BRONZE SPONSORS



Brad Bierbaum, Brian DeRevere and Ryan Peterson



Joe McKay and Mike Wolfe



# Underwriters and Other Donors

## UNDERWRITERS

Alabout Fire Inc.  
Ala Carte Catering, Inc.  
ALC Construction, LLC  
Berkeley Point Capital  
Calcrete Construction, Inc.  
CBRE, John Schumacher  
CBRE – Val Achtemeier,  
Darla Longo and Barbara Emmons  
Cell-Crete Corporation  
Champion Fire Systems, Inc.  
Chapman Glucksman Dean Roeb & Barger  
Colliers International,  
Steve Bellitti and Tom Taylor  
Cushman & Wakefield,  
Chuck Belden and Phil Lombardo  
DeRevere & Associates  
Dot Printer  
Fletcher Jones Motorcars  
FullScale Demolition, Inc.  
GuardTop, LLC  
Hendricks-Berkadia, Margie Molloy,  
Dean Zander & Vince Norris  
Integrated Services  
Interstate Restoration  
Invictus Construction Specialties  
J.P.Morgan Asset Management,  
Brian Okrent and Michael Yoo  
Link Murrel & Company, LLP  
MAC (Mechanical Air Control)  
Masterline Plumbing  
*Multihousing Professional*  
NASA Services, Inc.  
NOELLE Marketing Group  
*Orange County Business Journal*  
Partner Engineering & Science, Inc.

RealPage, Inc.  
Sherwin-Williams  
Silverline Construction Inc.  
Summit Plastering  
Traditional Jewelers  
Walker & Dunlop  
X-Act Finish & Trim, Inc.

## LIVE AUCTION

Antonello Ristorante  
Ben's Asphalt  
David Bonfadini  
Change a Life Foundation, Lisa Fujimoto  
JT Schmid's  
Kimco, Carolyne and Kim Megonigal  
Laura Khouri and Michael Hayde  
Majestic Realty, Ed Roski, Jr.  
Gary Peterson,  
Automatic Fire Sprinklers, Inc.  
Mary and Peter Rooney  
San Diego Chargers, Ken Derrett  
SRG Multifamily Acquisitions Group  
Nancy and Jeff Stack

## SILENT AUCTION

Aliso Viejo Country Club  
Arroyo Trabuco Golf Club  
Debbie Beers  
Steve Bellitti, Colliers International  
The Boulders, A Waldorf Astoria Resort  
Canali Men's Store  
Vince Ciavarella  
Datum Architects, Tim Lux  
Disneyland  
Fleming's  
Tom Gilchriese, TLG Paving  
House of Balsamic  
Island Hotel Newport Beach  
Janet Keller and Bernie Schneider  
Samra and Bill Keller  
Mike Kerr  
Kris Kristjansson  
Levendi Winery  
L'Occitane  
Lawrence Malekow,  
Howard Roofing Company, Inc.  
Janine Padia  
Paper Source  
Paradise Pen



Pelican Hill  
RL and David Peters  
Margaux Pierog  
Quiksilver  
Tim Quinn  
Mike Reehl  
Zoe and Don Solsby  
South Coast Plaza  
Nancy and Jeff Stack  
Studio 4 Salon  
Tijeras Creek Golf Club  
Tinder Box International  
Virginia Country Club, Jamie Mulligan  
Tracy and Mike Wood

## SUPER RAFFLE

Balboa Bay Resort  
MAC (Mechanical Air Control)  
Nirve Sports, Ltd.

## RAFFLE

Angels Baseball LP  
The Capital Grille  
Mary and Peter Rooney  
SRG Commercial Property  
Services Group  
Nancy and Jeff Stack

## GOODIE BAGS & GIVEAWAYS

The Catalano Family  
Contempo Creations  
Independence Bank  
Integrated Services  
Quiksilver  
Nancy and Jeff Stack  
The Wine Club, Inc.,  
Ron Van Vlaardingen





# Many Hands Make Lite Work

TOGETHER WE ARE MAKING A DIFFERENCE



*Lars and his mom Ashton Jenkins with family friend Mary Ann Franson.*

Mary Ann Franson and her family, dear friends to the Jenkins family, were kind to organize a huge yard sale to support Sam and Lars, and the event was a tremendous success!

The group raised \$3,328 for cystinosis research. The love and support of our community has truly lifted our spirits.

We feel motivated to continue our fundraising efforts. Not only is it helping cystinosis research, but it is also about bringing communities together.

*By Ashton Jenkins*



## Memorial Golf Tournament

The Second Annual Fore Fathers Memorial Golf Tournament was held on August 10, at the Boulder Creek Golf Course in Langdon, Alberta, Canada. The sold-out event was organized in honor of four fathers: John McCullagh, Conway Cameron, Frank Halluk and Gordon Cunningham, all who passed away in their sixties from heart-related illnesses.

Karen McCullagh and her brothers hosted the event to support two great causes: The Heart & Stroke Foundation, and Cystinosis Awareness & Research Effort (CARE), in honor of Andrew Cunningham, Karen's 10-year-old son, who was diagnosed with cystinosis as an infant. Andrew is the grandson of John McCullagh and Gordon Cunningham. The event raised nearly \$27,000, to be split between the two organizations.

Congratulations and thanks to everyone who helped create a lasting legacy for four fathers, and provide hope for a future without cystinosis.



*Andrew Cunningham with his sister Kelsey*



**ONLINE AUCTION**



Joshua's Journey of Hope held its online auction from October 1–10 on the *Bidding for Good* all-charity website. More than 200 businesses, such as Disneyland, Sea World, San Diego Zoo, Magic Mountain and numerous restaurants contributed items.



Items included an elephant clock, a Waterford decanter, gift items, collectibles and admission to attractions. Buyers came from many states, allowing us to receive 3 or 4 bids per item. With money still coming in, we may break the \$10,000 mark.

Brochures explaining the disease and thank you notes were included with all items. Joshua's foundation is very small, so we are extremely pleased with the success of this year's auction and our fundraising efforts to help find a cure for cystinosis.



## Golf Classic Raises Awareness for Cystinosis

By Tiffany Wilson, originally published at [www.shorelinebeacon.com](http://www.shorelinebeacon.com)

Sunday, September 22, 2013 was a day to remember for friends, families and athletes.

The Saugeen Shores Winterhawks, in collaboration with the Little family, held a *Swing, Shoot and Liv* golf classic at Saugeen Golf Club in support of cystinosis research and the Winterhawks. The event, the first of many fundraising efforts, raised approximately \$20,000 for the cause.

“We had a great turnout and great support from our community,” said Erin Little, mother of Olivia Little, who was diagnosed with cystinosis. “We went from 11 teams registered two weeks ago, to a total of 34 registered today.”

Olivia, who is three-and-a-half-years-old, was diagnosed with cystinosis in 2011. It is a rare, inherited disease

*Chad Little helps three-and-a-half-year-old daughter Olivia as she tees-off on the first hole to kick-start the afternoon full of golf.*

characterized by the abnormal accumulation of amino acid cystine. The buildup of cystine in the cells eventually destroys all major organs of the body.

“With such a rare disease affecting such a small population, research money is scarce to nonexistent,” Erin added.

To kick-off the Winterhawks and Little’s fundraising efforts, the golf tournament saw 136 golfers take to the links for a round of 18 holes, a barbecue lunch, full-service dinner and a chance to win great prizes.

“We want to continue to raise money because research is critical,” Erin said as she tried fighting back tears. “We need to raise funds to make a difference and raise awareness so this never happens again.”

---

## Love at First Sight



Victoria was our babysitter for 10 years, beginning when she was only 16 years old. She came into our lives before Holt was even conceived and has bravely faced this cystinosis challenge with us head on. Not surprisingly, I refer to her as my “angel straight from Heaven.” In September, Victoria got married to a wonderful young man name Zack Gadberry in Asheville, North Carolina. As part of their wedding celebration, Victoria and Zack had cards printed for each place setting at their wedding dinner letting guests know that in lieu of wedding favors, the bride and groom donated what would have been the cost to the Hope for Holt Foundation.

She and Zack have also donated a six-course dinner for 10 people complete with wine pairings each year for our Hearts for Holt live auction. It has quickly become one of our hottest and most sought after items.

Thank you Victoria and Zack for all your love and support. We couldn’t do it without you. *By Chrissy Grier, Holt Grier’s mom.*



## Wet Tee Shot

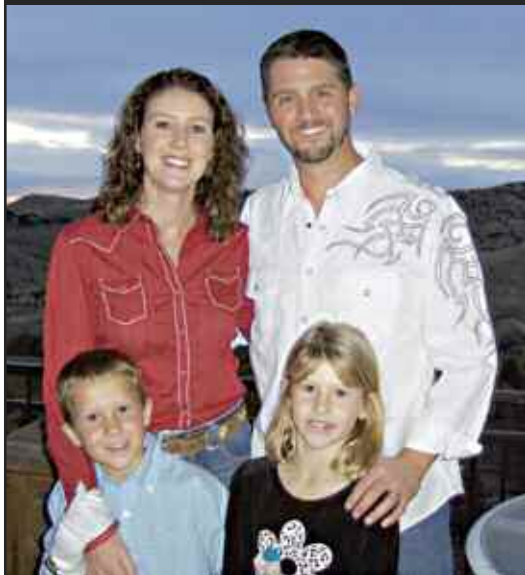
The Pacific Empire Radio held its *Wet Tee Shot* event at the Lewiston Golf & Country Club for five weeks in May and June. Golfers had a chance to win up to \$10,000 in cash and prizes.

For the third year in a row, Tina's Hope for a Cure was the chosen charity. The event, which is always great fun, raised \$761, with every dollar going directly to cystinosis research.

Thank you to Pacific Empire Radio, event sponsors and participants, for helping to make the event such a success, and for helping us find a cure for cystinosis!



*Mark, Denice and Tina Flerchinger, all at left, with DJs Don Kelly, standing, Evan Yeoman, kneeling, and Mark Bone, sitting, at the Wet Tee Shot event.*



## DEAR FRIENDS MAKING A DIFFERENCE

Since Jenna's and Patrick's diagnosis in 2005, the Lund Family has generously given \$1,000 each year. This amazing donation has been matched dollar for dollar by Hewlett Packard, Aunick's employer.



*The Ramirez brothers, (l-r) Elias, 12; Carlos, 14; and Andres, 16.*

## YOUNG MEN WITH LOTS OF HEART

The Ramirez brothers are pretty typical boys in many ways. Andres, 16, and his brothers, Carlos, 14, and Elias, 12, love rugby, soccer and football. What sets them far apart from many other teens and near-teens is their big hearts.

The three brothers, who have known the Partington twins since they were infants, have developed real respect for Jenna and Patrick's exceptional resiliency and love of life. The high school junior, freshman, and seventh grader have also stepped forward to make their respect tangible. Over the past five years the Ramirez brothers have given hundreds of their hard-earned allowance dollars to Jenna and Patrick's Foundation of Hope to support cystinosis research.

**Congratulations guys. You may be young but you're wonderful role models for "kids" of all ages.**



*Patrick Partington with special friend, 16-year-old Andres Ramirez.*





# IT TAKES A VILLAGE

## WITH LOTS OF ACTIVITIES

**Sunday, December 8, 2013**

*Race for Nicole – Dallas Marathon in honor of Nicole Hall*  
Dallas Marathon, Dallas, Texas  
Contact Aaron Hall at [Aaron\\_j\\_hall@yahoo.com](mailto:Aaron_j_hall@yahoo.com)



**Saturday, January 25, 2014**

*Seventh Annual Hearts for Holt Event – Holt Grier*  
Charlotte Country Club, North Carolina  
For tickets or to donate items visit [www.hopeforholt.org](http://www.hopeforholt.org)



**Friday, February 21, 2014**

*Jenna & Patrick's Foundation of Hope Fourth Annual Swing Golf tournament*  
Teal Bend Golf Course, Sacramento, California



**Saturday, February 22**

*Jenna & Patrick's Foundation of Hope Bling Gala event*  
The Citizen Hotel – Sacramento, California  
For tickets and information visit [www.jennaandpatrick.org/news-events/](http://www.jennaandpatrick.org/news-events/)



CYSTINOSIS RESEARCH FOUNDATION |  FOURTH INTERNATIONAL Cystinosis Research Symposium

**Thursday, March 6 – Friday, March 7, 2014**

*CRF International Cystinosis Research Symposium*  
The Beckman Center of the National Academies of Science and Engineering in Irvine, California  
Contact Nancy Stack at [nstack@cystinosisresearch.org](mailto:nstack@cystinosisresearch.org)

**Friday, March 21 – Saturday, March 22, 2014**

*Sixth Annual 24 Hours of Schweitzer – Henry Sturgis*  
Schweitzer Mountain Resort, Sandpoint, Idaho  
Registration opens December 1, 2013.  
Visit [www.24hoursforhank.org/events.html](http://www.24hoursforhank.org/events.html)



**Saturday, March 22, 2014**

*Music for Mary Benefit Concert – Mary Head*  
Tacoma Sportsman's Club, Puyallup, Washington  
For information contact Melissa Head at [mhead997@gmail.com](mailto:mhead997@gmail.com)



**Thursday, April 3 – Saturday, April 5, 2014**

*Cystinosis Research Foundation – Day of Hope Family Conference*  
Balboa Bay Club, Newport Beach, California  
For more information contact Nancy Stack at [nstack@cystinosisresearch.org](mailto:nstack@cystinosisresearch.org)



**Saturday, April 5, 2014**

*Cystinosis Research Foundation Natalie's Wish Event*  
Balboa Bay Club, Newport Beach, California  
For more information contact Zoe Solsby at [zsolsby@cystinosisresearch.org](mailto:zsolsby@cystinosisresearch.org)

**Saturday, May 17, 2014**

*Sixth Annual Wine, Stein and Dine Event – Tina Flerchinger*  
Rogers Toyota Scion Showroom, Lewiston, Idaho  
For information contact Denice Flerchinger at [mdflerch@gmail.com](mailto:mdflerch@gmail.com)



# CYSTINOSIS PATIENT RESOURCES

This resource list has been compiled by the Cystinosis Research Foundation for cystinosis patients and their families as a quick reference to assist you with your questions concerning insurance, and the process necessary to obtain medications and prescriptions. This information is intended for general education and should not be construed as advising on diagnosis or treatment of this or any other medical condition.

## PROCYSBI™

[www.procysbi.com/](http://www.procysbi.com/)

PROCYSBI™, approved by the FDA on April 30, 2013, is the first new cystine-depleting medication for nephropathic cystinosis in nearly 20 years.

PROCYSBI™ works by continuously reducing the toxic accumulation of cystine, and it can provide continuous control of cystine levels when taken every 12 hours and plays an important role in the management of nephropathic cystinosis.

Raptor Pharmaceuticals is the distributor of the PROCYSBI™ medication.

Patients must make an appointment with their doctor to determine if PROCYSBI™ is right for them. **Patients are strongly encouraged to enroll in RaptorCares™, a patient support program, created by Raptor Pharmaceuticals in preparation for their doctor's visit regarding PROCYSBI™ prescriptions.**

Enroll at [www.RaptorCares.com/QA3](http://www.RaptorCares.com/QA3) or call 855-888-4004.

## RAPTORCARES™

[www.RaptorCares.com](http://www.RaptorCares.com)

RaptorCares™ is a patient-support program, created by Raptor Pharmaceuticals, with input from nephropathic cystinosis patients and caregivers who understand the challenges and successes you may experience.

RaptorCares™ offers important information about nephropathic cystinosis as well as valuable tools to help you better manage the challenges along the way.

## CYSTAGON®

Cystagon® capsules contain cysteamine bitartrate, a cystine depleting agent that lowers the cystine content of cells in patients with cystinosis.

**CVS ProCare is the sole distributor for Cystagon®**

**Contact CVS ProCare:**

[ProCare.AD@cvsprocare.com](mailto:ProCare.AD@cvsprocare.com)  
888-700-0024

Hours of Operation (EST):

Monday – Friday: 8 a.m. – 8 p.m.

Saturday: 8 a.m. – 1 p.m.

**CVS ProCare provides**

**the following services:**

Counseling on ordering Cystagon®

Ordering other medications

Insurance inquiries

Trained pharmacist assistance  
available 24 hours

## CYSTARAN™ (EYE DROPS)

[www.cystaran.com](http://www.cystaran.com)

CYSTARAN™ is the only FDA-approved ophthalmic therapy for corneal crystals in cystinosis patients.

For information about CYSTARAN™ call Accredo Specialty Pharmacy at: 800-440-0473.

To contact Sigma-Tau, call Lesli King, Senior Manager, Patient Affairs: 301-670-5450 or [lesli.king@sigmatau.com](mailto:lesli.king@sigmatau.com).

Prescribing information is available at [www.cystaran.com](http://www.cystaran.com)

## NIH: NATIONAL INSTITUTES OF HEALTH

[www.nih.gov](http://www.nih.gov)

**Appointments with Dr. William Gahl**

**Contact:**

Joy C. Bryant, RN, BSN, CCRC

Research Nurse Specialist

301-443-8690, 102-10460 (page)

National Human Genome Research Institute

National Institutes of Health

9000 Rockville Pike

Building 10/Room 3-2551

Bethesda, Maryland 20892

## CYSTINOSIS RESEARCH FOUNDATION (CRF)

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

The Cystinosis Research Foundation is dedicated to finding better treatments to improve the quality of life for those with cystinosis and to ultimately find a cure for this devastating disease.

The Cystinosis Research Foundation issues grants for bench and clinical research studies bi-annually in order to accelerate research progress and ensure that cystinosis research is on-going and focused on novel treatments and a cure.

The Cystinosis Research Foundation is also dedicated to educating the public and the medical community about cystinosis to ensure early diagnosis and proper treatment.

Cystinosis Research Foundation  
18802 Bardeen Avenue, Irvine, CA 92612  
949-223-7610

## CCIR: CURE CYSTINOSIS INTERNATIONAL REGISTRY

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

CCIR is a collaborative effort by the leaders in the cystinosis community to identify people with cystinosis worldwide, to collect their medical history and information, and to share their de-identified (anonymous) information with the research community in an effort to accelerate novel treatments and a cure for cystinosis.

### For questions contact:

Betty Cabrera, CCIR Curator  
858-822-3747

email: [curator@cystinosisregistry.org](mailto:curator@cystinosisregistry.org)

## CCIR PARTNERS AND ADVOCATES

### Cystinosis Research Foundation

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

### Cystinosis Foundation

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

### Cystinosis UK

[www.cystinosis.org.uk](http://www.cystinosis.org.uk)

### Cystinosis Australia

[www.cystinosis.com.au/about.html](http://www.cystinosis.com.au/about.html)

### Cystinosis Foundation Germany

[www.cystinose-selbsthilfe.de](http://www.cystinose-selbsthilfe.de)

### Cystinosis Foundation Ireland

[www.cystinosis.ie](http://www.cystinosis.ie)

### Cystinosis Mexico AC

### Cystinosis Support Group South Africa

[www.cystinosis.co.za](http://www.cystinosis.co.za)

### Cystinosis Awareness & Research Effort

[www.cystinosis.ca](http://www.cystinosis.ca)

### Tina's Hope for a Cure

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

### Joshua's Journey of Hope

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

### Jenna and Patrick's Foundation of Hope

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

### Hope for Holt

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

### 24 Hours for Hank

[www.24hoursforhank.org](http://www.24hoursforhank.org)

## NORD: NATIONAL ORGANIZATION FOR RARE DISORDERS

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

NORD's vision and guiding principles on which our advocacy initiatives are based:

- A national awareness and recognition of the challenges faced by people living with rare diseases and the associated costs to society.
- A nation where people with rare diseases can secure access to diagnostics and therapies that extend and improve their lives.
- A social, political and financial culture of innovation that supports both the basic and translational research necessary to create diagnostic tests and therapies for all rare disorders.
- A regulatory environment that encourages development and timely approval of safe and effective diagnostics and treatments for patients with rare diseases.

## MEDICAL RESOURCES

### Gene Tests

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

### Clinical Genetic Testing

- [www.cincinnatichildrens.org/service/s/star/genetic-testing/](http://www.cincinnatichildrens.org/service/s/star/genetic-testing/)  
Test must be ordered by a physician
- University of California, Irvine – Mitomed Diagnostic Laboratory  
[www.mammag.uci.edu/mitomed](http://www.mammag.uci.edu/mitomed)
- University of California, San Diego  
[www.cystinosiscentral.org](http://www.cystinosiscentral.org)

### Kidney Transplants and Donation

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

## OTHER RESOURCES

### CheckOrphan

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

### Genetic Alliance

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

- U.S. Department of Health & Human Services  
National Health Information Center  
[www.Healthfinder.gov](http://www.Healthfinder.gov)

## RARE: THE GLOBAL GENES PROJECT

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

The Global Genes Project is one of the leading rare and genetic disease patient advocacy organizations in the world. The non-profit organization is led by Team RARE and promotes the needs of the rare and genetic disease community under a unifying symbol of hope – the Blue Denim Genes Ribbon™.

Rare and genetic diseases affect 1 in 10 Americans, 30 million people in the United States, and 300 million people globally. Over 7,000 distinct rare diseases exist and approximately 80 percent are caused by faulty genes. The National Institutes of Health estimates that 50 percent of people affected by rare diseases are children, making rare diseases one of the most deadly and debilitating for children worldwide.

It is estimated that 95 percent of all rare diseases do not have a single FDA-approved drug treatment, and there are currently fewer than 400 treatments approved by the FDA for rare diseases.

According to NIH estimates it will take 10,000 years at the current rate of FDA approvals to find therapies for all people suffering from rare and genetic diseases.



**CYSTINOSIS**  
RESEARCH FOUNDATION

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

# MATCHING GIFTS REALLY HELP

Corporations establish Matching Gift Programs to support their employees' charitable efforts to eligible nonprofit organizations.

## THE GREAT TREASURE HUNT!



**Exxon Mobil, Apple, Google, Microsoft, Berkshire Hathaway, Wal-Mart, Johnson & Johnson, Chevron, General Electric and Wells Fargo**

These are ten of the largest companies in the world, and they well could be the source of rich and unexpected treasures in our quest for the cure for cystinosis.

Each of these companies, with thousands of others, representing millions of people, offer Matching Gift Programs. In fact, more than 65 percent of Fortune 500 companies and thousands of smaller ones also offer them.

### A TREASURE TROVE OF UNTAPPED OPPORTUNITY

Greatnonprofits.org, a leading resource for nonprofit information, confirms that more than \$10 billion is left on the table annually through unused Corporate Matching Gift Programs. That's because many employees simply aren't aware of Corporate Matching Gifts. The information isn't always easy to locate, but it's worth the effort.

Fortunately, there are two easy ways to determine if your company offers such a program:

- First, review your employee manual or benefit book.
- Second, ask a representative in your HR department.

### WONDERING IF IT'S WORTH THE EFFORT FOR A SMALL GIFT? THE ANSWER IS YES!

There are countless examples of nonprofit organizations across the United States who received large gifts in a single year or over a period of time that originated from one or more of these programs.

Programs vary by company, but a typical match is dollar-for-dollar up to a set amount that is generally between \$2,000 – \$10,000 per employee each year. For example: if you work for Google and donate \$100 to the Cystinosis Research Foundation, Google will also send a check for \$100 to CRF, thereby doubling your gift.

### YOU CAN HELP AND IT'S EASY – FOR LARGE GIFTS AND SMALL ONES

There are countless ways to help the Cystinosis Research Foundation and its sister organizations each year. Now imagine how you'd feel if you could double your gift and your impact simply by filling out the proper paperwork with your employer.

Matching Gift Programs can have a significant impact on CRF's research efforts with no additional cost to you and with very little work on your part.

.....  
This is a partial list of corporations that offer matching gift programs:

- Abbott Laboratories
- AT&T
- Bank of America
- Boeing
- Capital Group Companies
- Cisco Systems
- Coca-Cola
- Duke Energy
- Fidelity Investments
- Gap Corporation
- GlaxoSmithKline
- Hewlett Packard
- Home Depot
- IBM
- Intel
- J.P. Morgan Chase & Co.
- John Deere
- Kaiser Permanente
- Kelloggs
- Kraft Foods
- McDonalds
- PG&E
- PepsiCo
- Pfizer
- PIMCO
- Qualcomm
- Starbucks
- Verizon
- Walt Disney
- Western Digital
- Yum Brands

If your employer isn't listed, there's still a good possibility that they offer a matching gifts program. Talk to your HR department for more information.

.....



**HIGHLY RESPECTED  
RESEARCHER  
TO KEYNOTE AT THE  
FOURTH INTERNATIONAL  
RESEARCH SYMPOSIUM**

**MARCH 6 AND MARCH 7, 2014  
Arnold and Mabel Beckman  
Center of the National Academies  
of Science and Engineering  
in Irvine, California**

**Cystinosis Research Foundation Board Chair Nancy Stack has announced that Ana Maria Cuervo, MD, PhD, will be the keynote speaker for the foundation's international symposium in March 2014. Dr. Cuervo is a renowned autophagy researcher from Albert Einstein College of Medicine in New York City.**

Corinne Antignac, MD, PhD, Hôpital Necker-Enfants Malades, Paris, France; Stéphanie Cherqui, PhD, University of California, San Diego; Julie Ingelfinger, MD, Harvard Medical School; and Elena Levchenko, MD, PhD, University Hospital Leuven, Belgium, have agreed to serve again as co-chairs for the upcoming conference.



*Ana Maria Cuervo, MD, PhD*

**Ana Maria Cuervo, MD, PhD will be the keynote speaker for the 2014 conference.** Dr. Cuervo is on the faculty at Albert Einstein College of Medicine in New York City.

She is co-director of the Einstein Institute for Aging Research, and a member of the Einstein Liver Research Center and Cancer Center. In 2001 she started her laboratory at Einstein, where she studies the role of protein-degradation in aging and age-related disorders, with emphasis in neurodegeneration.

Dr. Cuervo is a leader in the field of autophagy – the process by which cells recycle their waste. The Barcelona native is also an expert on the molecular biology of aging. Dr. Cuervo has been quoted in numerous publications, including *The New York Times* and *The Scientist*. She is co-editor-in-chief of *Aging Cell* and associate editor of *Autophagy* and has served on various NIH advisory panels and study sections.

The CRF's International Research Symposium is held every two years and is only open to invited researchers and clinicians. The approximately 65 invitees are experts in their fields, and on some aspect of the care and treatment of cystinosis. They are from leading international universities and research institutions.

The conference takes place on Thursday, March 6 and Friday, March 7, 2014, and will again be held at the Arnold and Mabel Beckman Center of the National Academies of Science and Engineering in Irvine, California.

The symposium has become known for the openness of the attendees, and for their willingness to share ideas and collaborate. Past attendees have commented that the conference fosters synergy, rare in the scientific community, which has played a key role in their individual and group success.



*Corinne Antignac, MD, PhD*



*Stéphanie Cherqui, PhD*



*Julie Ingelfinger, MD*



*Elena Levchenko, MD, PhD*

**For more information about the symposium, contact foundation Chair Nancy Stack at 949-223-7610 or [nstack@cystinosisresearch.org](mailto:nstack@cystinosisresearch.org).**

# CRF IN THE NEWS

Cystinosis families and their communities around the world are committed to finding a cure for cystinosis. Through their efforts information about the disease has spread and the resulting awareness of cystinosis has helped raise millions of dollars for research.

National, regional and local media outlets have also played a significant role in telling the story of the challenges those living with cystinosis face every day. Here is a partial sampling of stories that have appeared as we journey towards the cure.

## San Francisco Chronicle

Wednesday, May 8, 2013, Print and online editions

[www.sfchronicle.com/health/article/](http://www.sfchronicle.com/health/article/)

[New-drug-offers-hope-for-cystinosis-4497108.php](http://New-drug-offers-hope-for-cystinosis-4497108.php)



## The New York Times

Wednesday, May 1, 2013, Print and online editions

[www.nytimes.com/2013/05/01/business/fda-approves-raptor-drug-for-form-of-cystinosis.html?\\_r=0](http://www.nytimes.com/2013/05/01/business/fda-approves-raptor-drug-for-form-of-cystinosis.html?_r=0)

Andrew Pollack has covered the business and science of biotechnology at *The New York Times* since 2000. In April of this year, he interviewed Nancy and Natalie Stack for an article about cystinosis, Procysbi™, and the cost and challenges of bringing the drug to market. Pollack's article appeared in the print and online versions of the paper on May 1, 2013.



FDA approval of Procysbi™ in April 2013, spurred a number of media stories, including one in the *San Francisco Chronicle* that featured Jenna and Patrick Partington from Sacramento.

## ORANGE COUNTY REGISTER OC Register Magazine

June 25, 2013

### THE OC WISH HEARD ROUND THE WORLD

Cover story with eight pages featuring Natalie Stack and her life with cystinosis as she matures from an infant with a rare disease into a young college graduate who is about to enter graduate school and her chosen profession.



# Alaska Airlines

MAGAZINE

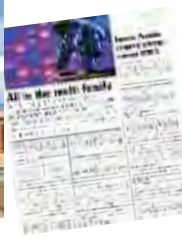
November 2013



## MULTIHOUSING PRO

magazine

August 2012



## ORANGE COUNTY BUSINESS JOURNAL

January 2013



TRADITIONAL Jewelers Magazine  
Spring/Summer 2013



Here are a few examples of press coverage CRF and the cystinosis community have received so far in 2013:

## NewportBeach-CoronadelMar Patch

### CDM GIRL'S BIRTHDAY WISH SPURS 'LIFE-SAVING' MEDICINE

Online May 3

<http://newportbeach.patch.com/groups/business-news/p/corona-del-mar-mom-nancy-stack-applauds-fda-approval-3aec8f45a7>



Nancy and Natalie Stack

## LEWISTON TRIBUNE

### FUNDRAISER WILL HELP FAMILY FIGHT RARE DISEASE – TINA'S HOPE FOR A CURE

May 18

[http://lmtribune.com/northwest/article\\_830cbf72-1ef6-561c-8c6b-5ea9bf50dde5.html](http://lmtribune.com/northwest/article_830cbf72-1ef6-561c-8c6b-5ea9bf50dde5.html)



### LC VALLEY SUPPORTS

#### "TINA'S HOPE FOR A CURE" EVENT

Lewiston, Idaho, May 22

[www.klewtv.com/news/local/](http://www.klewtv.com/news/local/Tinas-Hope-For-A-Cure-208563681.html)

[Tinas-Hope-For-A-Cure-208563681.html](http://www.klewtv.com/news/local/Tinas-Hope-For-A-Cure-208563681.html)

## FULLERTON TRIBUNE

### ONE MOM'S FIGHT TO SAVE HER SON

October 30, 2013

[www.ocregister.com/articles/](http://www.ocregister.com/articles/bailey-533822-dedio-kidney.html)

[bailey-533822-dedio-kidney.html](http://www.ocregister.com/articles/bailey-533822-dedio-kidney.html)

*Bailey DeDio and his mom Jessica DeDio celebrate at the first annual Bailey Believes Ride for a Cure in 2012. The second Bailey Believes Ride was held on November 9 in Barstow.*



## NORTH CAROLINA HEALTH NEWS

### NEW DRUG EASES LIFE FOR CHILD WITH RARE DISEASE

May 7, 2013

[www.northcarolinahealthnews.org/2013/05/07/new-drug-eases-life-for-child-with-rare-disease](http://www.northcarolinahealthnews.org/2013/05/07/new-drug-eases-life-for-child-with-rare-disease)

*Addison Cox and her mother, Nicole Cox at an event raising awareness for cystinosis.*



# KEEPING TRACK OF CYSTINE



A special blood test can give you important information.

Cystinosis is a challenging condition that can cause serious damage to your body.<sup>1,2</sup> One of the ways to prevent the damage is to keep your cystine levels as low as possible.<sup>3</sup> But how do you know what your cystine levels are? For that, you need to have a special white blood cell (WBC) cystine test.

## Why should I get tested?

In cystinosis, toxic levels of cystine can build up throughout the body.<sup>1</sup> If you delay taking your cystinosis medicine for even short periods of time, the cystine can build up.<sup>1</sup> The WBC cystine test tells you what your cystine levels are by measuring how much cystine has built up in your white blood cells.<sup>4</sup> Getting regular WBC cystine tests can help you stay on top of how your cystinosis medicine is working and how your body is responding to it.<sup>5</sup>

Learn more about  
WBC cystine testing  
and other information  
about cystinosis.  
Visit [KnowCystinosis.com](http://KnowCystinosis.com)

People who don't have cystinosis normally have cystine levels of about 0.2 nmol  $\frac{1}{2}$  cystine/mg protein.<sup>6</sup> People with cystinosis

can have up to 100 times the normal amount.<sup>7</sup> Many doctors say it's a good idea to get your cystine levels to less than 1 nmol  $\frac{1}{2}$  cystine/mg protein.<sup>7</sup>

## When should I have the test?

It's important to have the WBC cystine test done at the right time—based on your doctor's advice.

Your doctor may recommend having a WBC cystine test at least every 3 to 4 months.<sup>8</sup> That way, results can be compared to get a clearer picture of how your medicines are working over time. Your doctor can talk with you about what your levels should be and may adjust your medicines as needed.<sup>4</sup>

## How is the WBC cystine test done?

A sample of your blood is collected and prepared with a specific process to ensure accurate results. Then the blood sample is sent to another laboratory to be analyzed. When the results are ready, your doctor will discuss them with you.



At your next appointment, ask your doctor about WBC cystine tests and how often you should have them. You can work together to help you stay healthy—even with cystinosis.

**References:** 1. Leytchenko EN, van Doel CM, de Graaf-Hess AC, et al. Strict cysteamine dose regimen is required to prevent nocturnal cystine accumulation in cystinosis. *Pediatr Nephrol*. 2006;21:110-113. 2. Supportive treatment. Raptor Pharmaceuticals. <http://knowcystinosis.com/management-of-cystinosis/supportive-treatment>. Accessed August 22, 2013. 3. Gahl WA, Thoene JG, Schneider JA. Cystinosis. *N Engl J Med*. 2002;347(2):111-121. 4. Dalton N. The importance of accurate cystine level testing. [https://cystinosis.org/images/research/article-library/cystagon/2009\\_01\\_Cystine\\_Level\\_Testing.pdf](https://cystinosis.org/images/research/article-library/cystagon/2009_01_Cystine_Level_Testing.pdf). Published January 2009. Accessed September 9, 2013. 5. What are blood tests? National Heart, Lung, and Blood Institute. [http://www.nlm.nih.gov/health/dic/Diseases/bdl/bdt\\_what.html](http://www.nlm.nih.gov/health/dic/Diseases/bdl/bdt_what.html). Published January 6, 2012. Accessed August 22, 2013. 6. Goodyer P. The history of cystinosis: lessons for clinical management. *Int J Nephrol*. 2011;2011:929456. doi:10.4061/2011/929456. 7. Bellina EB, Huang MY, Schneider JA, Brundage RC, Tracy TS. Steady-state pharmacokinetics and pharmacodynamics of cysteamine bitartrate in paediatric nephropathic cystinosis patients. *Br J Clin Pharmacol*. 2003;56:520-525. 8. Prescribing information, PROCYSSE.



# Lay Abstracts — Spring 2013 Grants

## Angela Ballantyne, PhD, Principal Investigator; Doris Trauner, MD, Co-Investigator

**Project Title:** Patient-Reported Outcome and Health-Related Quality of Life in Adults with Cystinosis: A Study Utilizing the NIH “PROMIS”

**Objective/Rationale:** This is a 1-year continuation of a prior CRF-funded study. The objectives of the proposed study are to (1) gather vital information on patient-reported outcome and quality of life in the ever-growing population of adults with cystinosis, and (2) gather information on the final illnesses and specific causes of death in adults who had cystinosis and passed away during the past 10 years. This information can then be used as one method to weigh risks vs. benefits of emerging therapies, such as stem-cell therapy.

**Project Description:** Study participants will be at least 50 adults (ages 18 and older) diagnosed with cystinosis. (The goal is to obtain as many adults as possible, ideally more than 50.) This age group has been selected in order to examine outcome and quality-of-life in these individuals now that they are surviving well into adulthood. This study will utilize a computerized online questionnaire, the *Patient-Reported Outcomes Measurement Information System (PROMIS)*. The PROMIS was developed as part of the NIH’s 21st century “Roadmap” for medical research,

and is well validated and normed. An additional subject group will be included as an ancillary part of this study. Participants will be at least 20 parents (or nearest living relative) of individuals with cystinosis who died within the past 10 years. Information on the patient’s final illness and cause of death will be gathered from parents/relatives.

**Relevance to the Understanding and/or Treatment of Cystinosis:**

Findings of the proposed study will contribute a quality-of-life dimension to the fund of intensive research needed to consider stem cell therapy as a viable treatment approach for cystinosis. Moreover, the data will serve as a baseline from which future treatments/clinical trials for adults with cystinosis can be launched.

**Anticipated Outcome:** The advent of renal transplantation and cysteamine therapy have served to extend the lifespan of individuals with cystinosis into adulthood, but they do not prevent the ultimate progression of the disease. For this reason, it is important to learn about the longer-term effects of cystinosis on quality-of-life in adults with the disease, as well as the final illnesses and causes of death in this population. This knowledge can then be used in the planning of and/or justification for emerging therapies and treatments for adults living with cystinosis.

## Daryl Okamura, MD, Principal Investigator

**Project Title:** Elucidating the role of aberrant macrophage activation in nephropathic cystinosis

**Objective/Rationale:** Despite the vast improvements cysteamine bitartrate has brought to the treatment of patients with cystinosis, kidney disease remains a significant clinical problem and is associated with early mortality. Several long-term follow up studies demonstrate that even patients who initiate cysteamine therapy early in life, the majority will develop end-stage kidney disease before the age of 15 years. Studies in our lab and others have demonstrated that macrophages play a major role in the generation of oxidative stress and in the relentless progression of kidney fibrosis.

**Project Description:** The goal of the proposed studies is to further extend our investigations from the previous funding period by defining the cystinosis-deficient (CTNS<sup>-/-</sup>) macrophage phenotype and determining the mechanisms involved in its activation during nephropathic cystinosis. Our overall hypothesis is that CTNS<sup>-/-</sup> macrophages are genetically programmed to execute a more aggressive injurious, fibrotic response to kidney injury that serves an essential

role in the pathogenesis of cystinosis-associated chronic kidney disease. Two specific aims are proposed to continue our investigation of this new mechanistic paradigm: (1) To determine the CTNS<sup>-/-</sup> macrophage phenotype in response to cytokine activation and the mechanisms that lead to its altered behavior; and (2) To investigate the functional impact of the CTNS<sup>-/-</sup> macrophage phenotype on regeneration and fibrosis after kidney injury.

**Relevance to the Understanding and/or Treatment of Cystinosis:**

The results of these studies should provide the foundation for translational research studies based on the use of novel molecular and cellular adjunctive therapies directed at macrophages to prevent the development of nephropathic cystinosis.

**Anticipated Outcome:** Based on our preliminary data, it is anticipated that the results of the proposed studies will clearly establish that the cystinosis-deficient macrophage plays a critical role as a perpetuator of pro-fibrotic pathways initiated by injured kidney tubular epithelial cells and together promote the progression of nephropathic cystinosis.

# Lay Abstracts — Spring 2013 Grants

## Stéphanie Cherqui, PhD, Principal Investigator

**Project Title:** Mechanism of bone marrow stem cell-mediated therapy in the mouse model of cystinosis

**Objective/Rationale:** Adult bone marrow stem cells are promising tools for the development of innovative strategies for tissue repair and regenerative medicine. More specifically, hematopoietic stem cells (HSC) are the most attractive stem cells because of their easy access and their clinical use for over 30 years. However, the fate of the HSC after transplantation and the mechanisms by which they could lead to tissue repair is still unknown. The goal of this project is to reveal the mechanism(s) by which *Ctns*-expressing HSC can significantly improve cystinosis.

**Project Description:** We developed a new mouse model for cystinosis, the *DsRed Ctns*<sup>-/-</sup> mice expressing the *DsRed* reporter gene (appear red by microscope analysis) and transplanted with GFP-expressing HSC (appear green by microscope analysis).

This model represents a new innovative tool to study bone marrow-derived cell tissue integration because of the abundant stem cell integration occurring in the *Ctns*<sup>-/-</sup> mice and because of the dual color that allow distinguishing the transplanted cells and the host cells. We are going to perform a phenotypic and genetic characterization of these different cell populations to understand the mechanism by which they lead to tissue cystine decrease and tissue preservation.

**Relevance to the Understanding and/or Treatment of Cystinosis:**

This work is important for the future clinical application of stem cell therapy for cystinosis.

**Anticipated Outcome:** We are expecting to reveal new insights in hematopoietic stem cell-mediated regenerative medicine that could extend their use to other diseases.

## Stéphanie Cherqui, PhD, Principal Investigator

**Project Title:** Pharmacology/Toxicology studies for gene-modified stem cell transplantation for cystinosis

**Objective/Rationale:** Cystinosis is an inherited disease involving the defect of the gene *CTNS*. It causes cystine, a naturally occurring protein degradation products in the cells, to accumulate to toxic levels in the body. This leads to cell death and tissue damage to the kidneys, liver, eyes, muscle and brain. There is a treatment available that delays, the onset of these health problems, but it does not cure the disease. Thus, there is a pressing need for a better treatment for cystinosis.

**Project Description:** The long-term goal of this project is to develop a new treatment for cystinosis using gene therapy. It requires that the patient's own bone marrow stem cells be isolated and modified in the laboratory through the introduction of a normal version of the *CTNS* gene. These modified cells are then transplanted back into the patients to create a reservoir of healthy stem cells in the bone marrow that can rescue the organs throughout the patient's lifetime.

Preliminary studies have been done in the mouse model of cystinosis, the *Ctns*<sup>-/-</sup> mice, which develop similar symptoms as the patients. We transplanted mice stem cells that had been modified in the laboratory through the introduction of a functional *CTNS* gene into the *Ctns*<sup>-/-</sup> mice. This treatment significantly improved the disease in this model.

**Relevance to the Understanding and/or Treatment of Cystinosis:**

This proposal represents the safety and efficacy studies required by the Food and Drug Administration (FDA) that will establish whether it is warranted to test this therapy in humans who have cystinosis. This includes testing the impact of cysteamine on the stem cell transplantation in order to determine if cysteamine will be able to be used by patients who got transplanted with stem cells. This also includes the collection of information related to the natural history of cystinosis for the design of the clinical trial.

**Anticipated Outcome:** At the end of these safety studies, we should be able to initiate a clinical trial for gene-modified stem cell transplantation for cystinosis.



## Paul Goodyer, MD, Principal Investigator

**Project Title:** Novel Genetic Strategies for Cystinosis

**Objective/Rationale:** Although cysteamine therapy has changed the natural history of cystinosis, kidney transplantation is eventually required in every patient and devastating non-renal complications may appear in young adults. One day it may be possible to treat cystinosis patients with their own stem cells after correcting the mutated CTNS gene, but CTNS mutations vary from one patient to another. Here we propose a range of molecular strategies to fix the CTNS gene in ways which address the individual mutations seen in our patients.


**Project Description:** Among French Canadians, a specific mistake in the genetic code tricks each cell into stopping synthesis of cystinosis protein before completion. We will test new drugs that allow the cell to overlook the French Canadian mutation and others like it. For patients in whom the CTNS gene has been deleted, we propose a method to insert the normal gene in an alternative site within the DNA. For those with small errors in the CTNS gene that disturb its normal function, we have

devised a third approach that cuts that DNA and then guides the cell's natural repair system to insert the proper genetic code. All three strategies will be developed using a unique panel of skin cells derived from our patients that harbor the full range of cystinosis mutations.

**Relevance to the Understanding and/or Treatment of Cystinosis:**

If we can show that novel drugs allow cells to overlook the common French Canadian mutation, oral therapy might then be attempted to restore activity of the mutated gene in this group of patients. Strategies to edit the CTNS gene in cultured cells assume that similar techniques can be applied for correcting the gene in each patient's stem cells in the future.

**Anticipated Outcome:** New drugs have been developed that allow the cell to overlook mistakes in the genetic code or use the cell's natural repair mechanisms to fix them. These therapies are being applied to other genetic diseases. In this proposal, we are trying to learn how they might be applied in an individualized way to the specific mutations that cause cystinosis.



## Sergio D. Catz, PhD (Mentor), Gennaro Napolitano (Fellow)

**Project Title:** Small molecule regulators of vesicular trafficking to enhance lysosomal exocytosis in cystinosis

**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 macromolecules 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. Here, 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) understand the mechanisms of defective LAMP2a downregulation and mislocalization in cystinotic cells and to understand the mechanism of defective translocation

of substrates for degradation into the lysosomal lumen.

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 LAMP2a localization and 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.

## Published Studies (since our spring 2013 issue)

“Cysteamine Modulates Oxidative Stress and Blocks Myofibroblast Activity in CKD” – Published September 2013, *JASN (Journal of American Society of Nephrology)* by Daryl Okamura, MD, Assistant Professor of Pediatrics, Seattle Children’s Research Institute, Seattle, Washington.



“Is Genetic Rescue of Cystinosis an Achievable Treatment Goal?” – Published July 16, 2013, *Nephrology Dialysis Transplantation: Oxford University Journals* by Dr. Stéphanie Cherqui, University of California, San Diego.



## CRF Science Report and Research Grant Updates

CRF research grant progress reports are published in the *Research* section on our website: [www.cystinosisresearch.org](http://www.cystinosisresearch.org).

As updates are received they appear in our monthly *Star Facts* e-newsletter.

## 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

### CHAIR

**Corinne Antignac, MD, PhD**  
Hôpital Necker-Enfants Malades  
Paris, France

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

**Pierre J. Courtoy, MD, PhD**  
Louvain University Medical School  
De Duve Institute  
Brussels, Belgium

**Allison A. Eddy, MD, FRCP(C)**  
BC Children’s Hospital  
Vancouver, BC, Canada

**Francesco Emma, MD**  
Bambino Gesù Children’s Hospital  
Rome, Italy

**Lisa M. Guay-Woodford, MD**  
Children’s National Medical Center  
Washington, DC

**Julie R. Ingelfinger, MD**  
Harvard Medical School  
Boston, Massachusetts

**Martin Konrad, MD**  
University Children’s Hospital  
Muenster, Germany

**Elena Levchenko, MD, PhD**  
University Hospital Leuven  
Leuven, Belgium

**William E. Smoyer, MD, FASN**  
Center for Clinical and  
Translational Research  
Ohio State University

## 2013 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 \$22 million, with every dollar raised going directly to cystinosis research.

The CRF announced its fall 2013 call for research proposals and fellowships in March. Applications were due October 25, 2013.

**Currently, the Cystinosis Research Foundation has \$1 million available for new research grants and fellowships.**

Visit [www.cystinosisresearch.org/For-Researchers](http://www.cystinosisresearch.org/For-Researchers) for details.



## 2013 Grants Funded

**Angela Ballantyne, PhD**, Principal Investigator  
University of California, San Diego

“Patient-Reported Outcome and Health-Related Quality of Life in Adults with Cystinosis: A Study Utilizing the NIH PROMIS”  
\$44,923 – 1-year grant (February 1, 2013 – January 31, 2014)

**Sergio Catz, PhD**, Mentor  
Gennaro Napolitano, PhD, Fellow  
The Scripps Research Institute, La Jolla, California  
“Small Molecule Regulators of Vesicular Trafficking to Enhance Lysosomal Exocytosis in Cystinosis”  
\$74,938 – 1-year grant (October 1, 2013 – September 30, 2014)

**Stéphanie Cherqui, PhD**, Principal Investigator  
University of California, San Diego  
“Mechanism of Bone Marrow Stem Cell-Mediated Therapy in the Mouse Model of Cystinosis”  
\$344,337 – 2-year grant (September 1, 2013 – August 31, 2015)

**Stéphanie Cherqui, PhD**, Principal Investigator  
University of California, San Diego  
“Pharmacology/Toxicology Studies for Gene-Modified Stem Cell Transplantation for Cystinosis”  
\$499,971 – 3-year grant (September 1, 2013 – August 31, 2016)

**Paul Goodyer, MD**, Mentor  
Emma Brasell, Research Fellow  
Montreal Children’s Hospital, Canada  
“Novel Therapeutic Strategies for Cystinosis”  
\$75,000 – 3-year grant (September 1, 2013 – August 31, 2016)

**Daryl Okamura, MD**  
Seattle Children’s Research Institute, Seattle, Washington  
“Elucidating the Role of Aberrant Macrophage Activation in Nephropathic Cystinosis”  
\$256,208 – 2-year grant (September 1, 2013 – August 31, 2015)

## 2013 Interim CRF Grant Awards

**Ranjan Dohil, MD**, Principal Investigator  
University of California, San Diego  
“Funding Support for Betty Cabrera’s Curatorship of the Cure Cystinosis International Registry”  
\$87,217 – 3-year grant

**Ghanashyam Acharya, PhD**  
Baylor College of Medicine, Houston, Texas  
“Delta Vision Deconvolution Imaging System”  
\$198,000

The Delta Vision Deconvolution Imaging System will be used to conduct in vivo studies to optimize the nanowafer drug delivery system. The data generated by the system will accelerate current nanowafer research with the goal of translating the research into a clinical trial for novel treatments for cystinosis within the next 1–2 years.

The Cystinosis Research Foundation utilizes a Scientific Review Board (SRB) comprised of leading international experts in the field of cystinosis. (See list above left.) 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 and advises the CRF on the scientific merits of each proposal.

The Cystinosis Research Foundation’s goal is to speed promising cystinosis research toward clinical trials. To that end, CRF prioritizes research that will lead to better treatments and a cure for cystinosis. The 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 415 registrants from 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](http://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.



Ronald Reagan UCLA Medical Center

# RESEARCH STUDY OF BONE MARROW TRANSPLANT

## FOR PATIENTS WITH CYSTINOSIS FROM MATCHED RELATED DONOR

A research study is being done by physicians at the Ronald Reagan UCLA Medical Center in Los Angeles to determine if blood stem cell transplants, with bone marrow from matched related donors, are an effective treatment that will slow down or prevent the progression of cystinosis. The study will also look into the risks associated with this treatment and if they are acceptable considering the potential benefit of avoiding the long-term consequences of cystinosis. Bone marrow transplant has never been used to treat cystinosis in humans, but recent studies in a mouse model of cystinosis show that this form of therapy, not only helps, but prevents disease progression in mice. In this experimental study, the most successful form of bone marrow transplant, HLA-matched related donor bone marrow transplantation, will be performed.

### Eligibility criteria for participating in the study includes:

- 1 A) Adults, ages 18 and older with nephropathic cystinosis with stable kidney function (defined as less than 20% change in creatinine clearance from prior 12 weeks) and one or more of the following: a) muscle weakness; b) swallowing difficulties; c) progressive visual loss; d) intestinal malabsorption.

or

- B) Children ages 13-17 years who do not tolerate or do not take cysteamine (defined by leukocyte cystine levels greater than 5 nmol half-cystine/mg protein for 2 consecutive time points at least 3 months apart during the prior 6 months or parental confirmation of patient intolerance) and worsening clinical manifestations as determined by a physician who is not an investigator on this study.
- 2 Patients must have a related bone marrow donor who is HLA-matched on 10 of 10 alleles.
- 3 Patients with adequate physical function as measured by:  
Pre-transplant tests of heart, lungs, kidneys, liver, and other organs and must not have a serious infection, be pregnant, or have undergone a prior stem cell transplant.

### CRF Cystinosis Gene Therapy Consortium Members

**Corinne Antignac, MD, PhD**

Hôpital Necker-Enfants Malades,  
Paris, France

**Stéphanie Cherqui, PhD**

University of California, San Diego

**William Gahl, MD, PhD**

National Institutes of Health,  
Bethesda, Maryland

**Donald B. Kohn, MD**

University of California, Los Angeles

**Theodore B. Moore, MD**

Mattel Children's Hospital  
University of California, Los Angeles

**Daniel R. Salomon, MD**

The Scripps Research Institute,  
La Jolla, California

**Jerry Schneider, MD**

Dean for Academic Affairs Emeritus,  
University of California, San Diego

**Nancy Stack**

Cystinosis Research Foundation,  
Irvine, California

## New Clinical Research Study

Research Study of Bone Marrow Transplant for  
Cystinosis Patients From Matched Related Donor

**UCLA**

**Health**

Ronald Reagan UCLA Medical Center

Interested subjects should ask their physician to contact Zoe Solsby at the Cystinosis Research Foundation for additional information. Telephone: 949.223.7610 or email: [zsolsby@cystinosisresearch.org](mailto:zsolsby@cystinosisresearch.org).

# Cystinosis Research Foundation



We are indebted to everyone who serves on a Cystinosis Research Foundation Board for their leadership, guidance and commitment to helping our children.

## BOARD OF TRUSTEES

### CHAIR

**Nancy J. Stack**  
Cystinosis Research Foundation

### VICE CHAIR

**Geoffrey L. Stack**  
SARES•REGIS Group

### TREASURER

**Donald L. Solsby**  
Independence Bank

### MEMBERS

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

**David Christensen**  
Louisville United  
Methodist Church

**Bruce Crair**  
Technology Transformation

**Kris K. Elftmann**  
Noelle Marketing Group

**Denice Flerchinger**  
Tina's Hope For A Cure

**Whitney Glaize**  
Cystinosis Advocate

**Jason K. Grier**  
Hope for Holt  
McAfee, Inc.

**John S. Hagestad**  
SARES•REGIS Group

**Michael K. Hayde**  
Western National Group

**John C. Manly, Esq.**  
Manly & Stewart

**Kevin Partington**  
Jenna & Patrick's  
Foundation of Hope  
Cushman & Wakefield

**Teresa Partington**  
Jenna & Patrick's  
Foundation of Hope

**Jody Strauss**  
Cystinosis Awareness  
& Research Effort

**Tricia Sturgis**  
24 Hours for Hank

## MEDICAL AND SCIENTIFIC ADVISORY BOARD

### Stéphanie Cherqui, PhD

University of California, San Diego  
San Diego, California

### Ranjan Dohil, MD

University of California, San Diego  
San Diego, California

### Allison A. Eddy, MD, FRCP(C)

BC Children's Hospital  
Vancouver, BC, Canada

### Robert Mak, MD

University of California, San Diego  
San Diego, California

### Jerry Schneider, MD

Dean for Academic Affairs Emeritus  
University of California, San Diego  
San Diego, California

### Jennifer Simpson, MD

University of California, Irvine  
Irvine, California

### Doris Trauner, MD

University of California, San Diego  
San Diego, California



Zoe R. Solsby, Vice President

## MISSION

The mission of the Cystinosis Research Foundation is to find better treatments and a cure for cystinosis by supporting bench, clinical and translational research. CRF has dedicated almost \$17 million in ten years to cystinosis research in an effort to find a cure.

## EDUCATION

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

# CYSTINOSIS RESEARCH FOUNDATION Global Outreach. Global Impact.



In **2003**  
CRF was Founded

*To have  
my disease  
go away forever*

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



**1,000,000+**

CRF-FUNDED RESEARCH ALSO OFFERS  
**HOPE TO MILLIONS**  
WHO SUFFER FROM OTHER RARE AND  
WELL-KNOWN DISEASES  
INCLUDING HUNTINGTON'S  
DISEASE AND NASH  
(FATTY LIVER DISEASE)



**100 PERCENT**

**OF EVERY DOLLAR DONATED  
GOES DIRECTLY TO  
CYSTINOSIS RESEARCH**

DURING THE FOLLOWING  
**10 YEARS**

AND WITH SUPPORT FROM ITS  
MANY FRIENDS, CRF HAS RAISED

**\$22 MILLION**

TO BRING NATALIE'S WISH CLOSER TO REALITY



*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.*

