

Cystinosis

MAGAZINE

SUMMER 2014 FOR FRIENDS AND SUPPORTERS OF THE CYSTINOSIS RESEARCH FOUNDATION



*Currently, there is
no cure for cystinosis but
There is hope.*



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

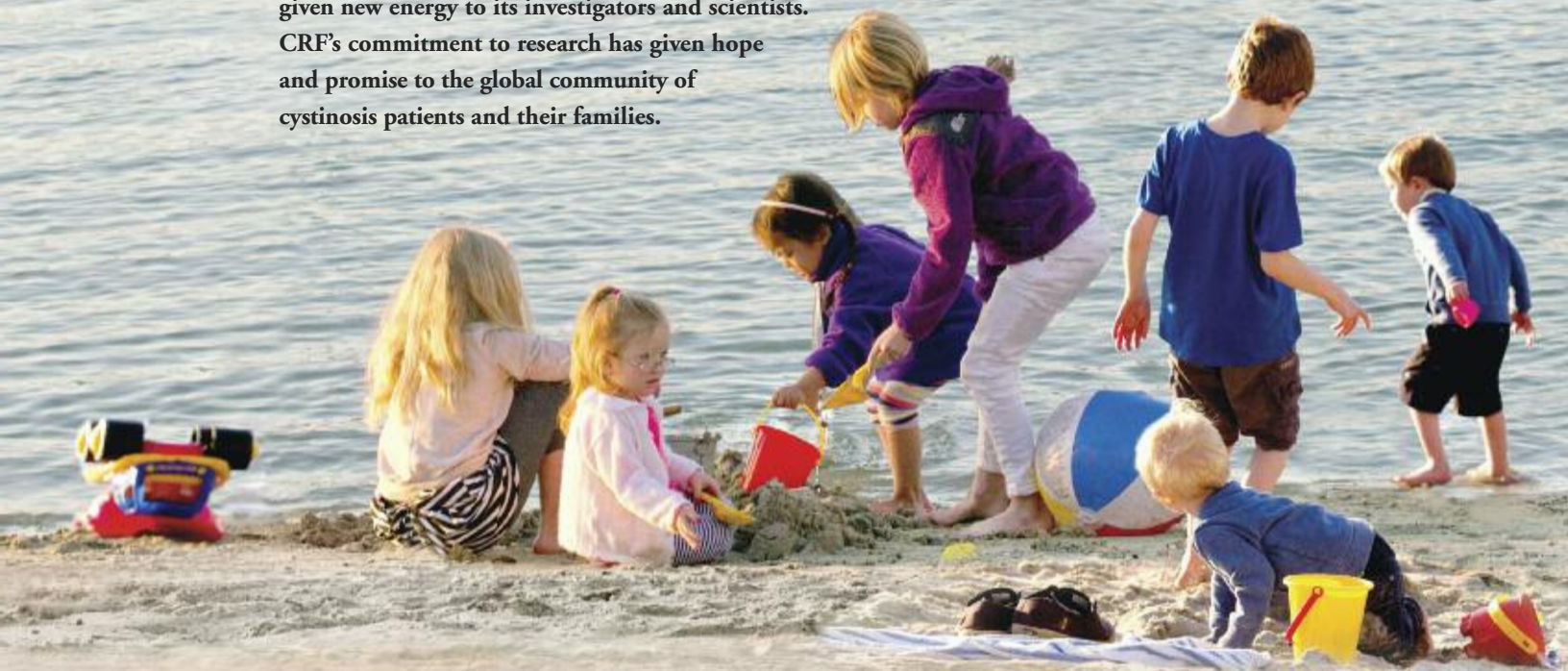
Medication is available to control some of the symptoms of this terrible disease, but cystinosis remains incurable.

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

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

The Cystinosis Research Foundation was established in 2003 with the sole purpose of raising funds to find better treatments and ultimately a cure for cystinosis. Today, CRF is the largest provider of grants for cystinosis research in the world, funding more than 114 studies in 11 countries. CRF has raised \$25.8 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.

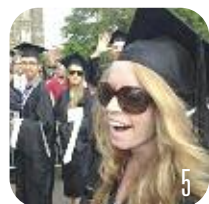
CRF's commitment to research has given hope and promise to the global community of cystinosis patients and their families.



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Cystinosis



On the cover:
 A quilt presented to CRF Board
 Chair Nancy Stack at the 2014
 Day of Hope Family Conference.
 See page 2 for more information.

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

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

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

THE YEAR OF THE QUILT

Dear Friends and Family:

I am certain that you are intrigued by the gorgeous quilt that graces the cover of this issue of *Cystinosis Magazine*. This quilt is particularly extraordinary because it is a community quilt, given to me by the cystinosis families and patients who are part of the CRF family.



Erin Little, the mother of Olivia who has cystinosis, is an artist with boundless talent and creativity. After seeing the CRF Day of Hope family conference logo, Erin was inspired to memorialize the logo (see page 4) by creating a quilt that involved the cystinosis community's participation. Erin asked families to send her a swatch of fabric so that she could include it in the creation of the quilt. Erin then spent countless hours stitching together all of the individual pieces of fabric to create the beautiful community quilt you see on the cover.

Only one word is sewn into the quilt – the word HOPE. Hope sustains us and allows us to dream of a life free of cystinosis. Hope is alive and powerful because we know that the research funded by CRF and supported by all of you brings us closer to a cure every day.

This quilt is symbolic of our community – its strength and its beauty, its colorfulness and its uniqueness. It was stitched together with love – each piece of fabric unique and distinct. It represents the connectedness of our community and the optimism we have that a cure will be found.

This unexpected gift is the most special gift ever given to me and I will cherish it forever. It is a reminder to me that we have more work to do to find the cure but along this journey, we are blessed to have each other.



THE REALITIES OF CYSTINOSIS

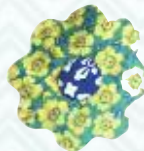
It is hard for me to believe that Natalie is 23 years old – it seems like only yesterday that I pushed her in a stroller, walked her in to her classroom for her first day of kindergarten, taught her how to drive, then watched her graduate from high school, and recently and proudly watched her graduate from college. As parents, we ask ourselves, where did the years go and why are they going by so quickly?

We are truly blessed that we have celebrated the milestones that parents plan and expect for their child but the milestones celebrated for Natalie have been in the shadow of the reality of cystinosis. We rejoice at Natalie’s milestones because we never forget that 23 years ago, we were told that Natalie had a progressive and fatal disease called cystinosis and that she would likely not live long enough to graduate from high school.

Cystinosis is a rare metabolic disease that affects every cell in the body. It eventually destroys every organ in the body including the liver, kidneys, eyes, muscles, thyroid and brain. On average our children take between **8-12 medications every day – Natalie takes 36 pills a day or 1,080 pills a month or 13,140 pills a year! And she takes eye drops 7-8 times a day to reduce her chances of going blind.**

Life with cystinosis is not easy. There are G-tube feedings, blood draws, growth hormone shots, kidney transplants, daily vomiting, and other gastrointestinal side effects. As our children live into adulthood, more complications occur including muscle wasting, difficulty swallowing, neurological issues and for some, blindness. The medications our children take cause constant discomfort and pain – cystinosis is a brutal disease.

RESEARCH PROGRESS EQUALS HOPE



CRF is the largest fund provider of cystinosis research in the world and we have made tremendous progress, yet cystinosis remains an incurable, progressive disease – we are close to the cure, but we are not there yet.

Since 2003, the year Natalie made her birthday wish, *to have my disease go away forever*, and the year the foundation began, what we know about cystinosis has changed dramatically. In 11 years, you have helped us create a thriving research community – we have issued 114 research grants in 11 countries and those researchers have published over 47 articles in prestigious journals. Because of your support, we have funded researchers whose work has exponentially increased the breadth and knowledge about cystinosis and as a result of that knowledge; new discoveries have been made about the pathogenesis of cystinosis.

It was CRF-funded doctors who discovered a delayed-release form of the critical, life-saving medication. That discovery allows our children to take their medication every 12 hours instead of every 6 hours, thereby increasing compliance and reducing the drug’s horrible side effects. Since its FDA approval last year, almost 300 children and adults with cystinosis are on the new medication – all because of you!



We are pleased to report that Dr. Stéphanie Cherqui, at the University of California, San Diego, is working with the FDA in anticipation of a clinical trial for a stem cell and gene therapy treatment. The treatment, if it works, will potentially cure cystinosis.

And on the horizon, is the astonishing nanotechnology work being done in collaboration by Dr. Jennifer Simpson at the University of California, Irvine and Dr. Ghanashyam Acharya at the Baylor School of Medicine. They are focused on a treatment for corneal cystinosis, the painful eye condition that causes severe photophobia and often blindness as they grow older. We anticipate a clinical trial for a novel treatment within a year.

Twenty three years ago, we had little hope for new treatments and hope for a cure eluded us. What a difference the last 11 years have made! Today, hope is abundant, which keeps us going as parents and patients. The hope we have today, is a direct result of the research.



The 2013 CRF Day of Hope logo on which the quilt was patterned



HELPING OTHER DISEASES

Many of you are aware that discoveries made by CRF-funded researchers are helping other diseases. Cystinosis research has helped advance potential treatments for more prevalent and well-known disorders and diseases such as Huntington's disease, Parkinson's disease and NASH – a progressive liver disease.

There is great potential that CRF stem cell research will help other corneal diseases and genetic diseases with systemic defects similar to cystinosis.

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

THE RESEARCH CYCLE MUST CONTINUE



Natalie is living at home for a few months before she starts the next chapter in her life. Now that she is home, I “see” cystinosis again. I observe the extreme fatigue she experiences as a result of cystinosis. She often says her eyes hurt and I can see how bloodshot they are because of corneal cystinosis. She has daily stomach aches and nausea because of the cocktail of medications she takes. Her dresser and nightstands are covered with bottles of pills – all the medications that keep her alive. Her routine is not “normal” but it is normal for her and normal for others with cystinosis. Having her home is a reminder that cystinosis is ever present and **there is no cure until we find it.**

I confess that I am nervous because I know that time is running out. I cannot forget that she has cystinosis and that it is a progressive disease. My faith is strong and my own determination steadfast – there is no other choice; we must work until we find a cure for our children.

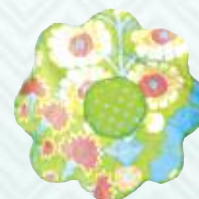
I dream that Natalie and the other children and adults with cystinosis will have a life free of cystinosis – no medications, no pain, no hospital visits and blood draws, no worries about life expectancy. I am certain that the research we fund will result in a cure so that every child and adult with cystinosis will someday be free of this disease.

Each and every one of you has supported us on this life journey. How can we possibly thank you for the gifts you have given us? Your unwavering support and prayers have lifted us up and given us the strength we need to continue the quest for the cure.

We have reached milestones and we are on the brink of new possibilities and treatments. With your support, we will continue to fund the best and the brightest researchers in the world. On behalf of all of the families with cystinosis, thank you for giving us the greatest gift of all – HOPE.

With blessings from our family to yours.

Nancy and Jeff



Dear Friends and Family:



I recently returned from my graduation ceremony at Georgetown University. It was a wonderful weekend spent with friends and family. I can't believe that I am done with my undergraduate work. I was notified in December that I was accepted to the USC School of Social Work graduate program! I was so excited – USC was my top choice for schools and it is a dream come true for me. I am not quite sure what my focus will be within the program but I know I would like to work with children. I want to impact the world in a positive way and a degree in social work will help me realize my dream of helping others. I currently live at home with my wonderful parents and just started a part-time job at a law firm.

I am so excited to start a new chapter in my life at USC. I am looking forward to meeting new people and exploring my passions there. Yet, as any new journey begins, it is always a little scary. For me, I fear I won't be able to get up early every morning because of extreme fatigue or a nauseous stomach from the medications I take every 12 hours. Although these are some of my worries, I can't let cystinosis be the reason I stop pursuing my dreams.

Cystinosis is a part of my life, everyday; it is hard and frustrating, but I know there are better treatments on the way and I know I have to live my life every day to the fullest. At the end of the day, it is MY life, and it is up to me to choose whether I want to see cystinosis as a burden or just a challenge to living an extraordinary life. I am strong BECAUSE of the obstacles cystinosis has challenged me with and I am optimistic BECAUSE I choose to live a positive and full life.

I am astonished at how far we've come since the day I made my wish 11 years ago. My friends and family have always supported me in every way they could and they never judged me or treated me differently.

The cystinosis community is small, but very BIG in so many ways. Our community has so much hope and dedication to finding the cure.

I am always amazed at how much love we give each other. My parents, the doctors and most importantly, the cystinosis community has made a life FREE of cystinosis more possible than ever before. I want to thank all of you for your generosity, and, most of all, your determination to make my wish come true.

I am so blessed to have such a supportive community of friends and families. With your love and support, everyone with cystinosis will continue to have hope for the cure.

Thank you for never giving up.

Love, Natalie



Day of Hope

CYSTINOSIS RESEARCH FOUNDATION FAMILY CONFERENCE

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

We just returned last night from the CRF Day of Hope Family Conference in Newport Beach, California. This was our second time as a whole family, and it is the highlight of the year, even bigger than Christmas!

nancy and Jeff Stack go all out to make the families feel welcome, comfortable, educated, and, most importantly, full of hope that life for people with cystinosis will get better and better. Forty families attended this year, and it was like going to a big family reunion.

Ashton and I chose to drive again because it's just easier for us to deal with screaming children in our own car rather than on a crowded airplane. It's also nice to have constant access to snacks (Lars's favorite word) and have room to bring blankets and sleeping pads and basically anything else we feel like cramming in our car. The boys made sure to bring their new stuffed animal dogs they got from Ashton's grandma, Jackie.

The conference kicked off with dinner at the Balboa Bay Resort. It's a fantastic evening where we see old friends and meet new ones while our children run wild (despite parental instructions). It's amazing to watch the kids together.

Sam reconnected with Henry Sturgis immediately, and also had fun playing with Jackson Blum and Andrew Cunningham, and chasing Tina Flerchinger. We were all pretty worn out that night from traveling, and went to bed right after medicine time.



On Friday morning we had breakfast and then dropped the kids off with the babysitters. This is one of the things we love the most. Lars was in heaven with the huge snack table and unlimited opportunities to scribble with markers. Sam loved playing with all his buddies and seeing Spiderman.

The conference started with all the families circling up, and we each got to get up and share what our wishes are for our loved ones with cystinosis. Ashton wished that Sam would be able to fulfill all his dreams, and that Lars would never need a kidney transplant. I wished that they would both have long and happy lives.

Sam wished that Lars would become a Greek soldier, and wished for himself to become a spy. We put our wishes on paper cutouts shaped like flowers and put them all up on a large tree representing our cystinosis family. It was really a beautiful symbol.

The best moment of the morning was when Kevin Partington lifted up his sleeve to show off the prototype patch that Dr. Ghanashyam Acharya has been working on for the last year as a better way to deliver cysteamine. Ashton and I both started crying when we saw that. There were a lot of tears shed in general that morning.

The rest of Friday we went to talks by the different researchers and physicians in the cystinosis community.

- ♥ Dr. Grimm gave a fantastic lecture on living with Fanconi syndrome.
- ♥ Dr. Mak got everyone excited about the potential benefits of Vitamin D on muscle wasting.
- ♥ Dr. Dohil described the research related to GI issues in cystinosis, including the development of Procsybi®.

♥ Dr. Jennifer Simpson and Dr. Ghanashyam Acharya gave updates on their nanowafer delivery system for corneal cystinosis, which should be in a clinical trial soon.

♥ Then Dr. Acharya spent just a few minutes talking about the development of the patch. Basically Ghanashyam invented a completely new model with multiple layers that can pump the cysteamine into the skin. He thinks he can get 500 mg, or half a gram, of cysteamine into the patch, and his goal would be to have it last three days.

Sam wielded his stick like a lightsaber, and instigated a war with the girls on the beach. He said Gabbie Strauss was the captain of the girls. It was pretty epic. We also sang happy birthday to Mack Maxwell, who turned 50 years old that night!

Saturday we resumed meetings and heard from Dr. Grimm again about kidney transplants, which was a very helpful and educational talk. Who knew that cats were so dangerous for transplant patients?! It's a good thing we're a dog family.



- ♥ Dr. Sergio Catz talked about a novel mechanism for improving removal of cystine from lysosomes, which involves trafficking by a protein called Rab27. He's looking for an existing drug that would induce Rab expression, forcing lysosomes to fuse with the cell membrane and dump their contents. It sounds like a pretty cool adjunctive therapy to enhance the effects of cysteamine.
- ♥ Dr. Cherqui gave us an update on her progress with the autologous stem cell transplant project, and said she hopes to have a clinical trial in 2–3 years.

All of these talks were followed by a question and answer session with the physicians and researchers, and then a brainstorming session for parents to discuss solutions to common problems like eating, bedwetting, etc.

Friday night was definitely Sam's favorite. We ate dinner on the private lawn/beach of the Balboa Bay Resort. Sam brought shovels and buckets and spent most of the evening digging and getting drenched. We finally had to drag him away from the water and change his entire outfit because he was completely soaked. There was cotton candy for the kids on plastic light-up sticks, which was a huge hit. The Stacks think of everything!

And it turns out cystinosis patients keep their transplants longer, probably because of the anti-scarring properties of cysteamine use. We also heard from Dr. Doris Trauner about neurologic issues in cystinosis, and Dr. Angela Ballantyne, who talked about practical ways of dealing with neurologic, behavioral and educational issues, as well as quality-of-life.

Then we heard from the real giants, the adult/teen panel. Rachel, Bailey, Joe, Jennifer, Shannon Keizer, Natalie, Tom, Mack, Bryan and Shannon Paju all shared inspiring insights about growing up, going to school, working and living with cystinosis. Some of the most interesting confessions surrounded medication compliance, which



Day of Hope

CYSTINOSIS RESEARCH FOUNDATION
FAMILY CONFERENCE

Please join us
April 16–18, 2015
for next year's
Family Conference

Newport Beach
California

*I loved every minute of
the conference start to finish.*



*I had a great time and every day
I remember something
else I learned.*



*This event re-energizes my hope
for a cure and refuels me
for another year!*



*The conference was amazing.
I can't wait to see you next year.*



*I don't know how we can ever
repay you for the opportunity to
come and learn more about this
disease and meet the families.*

Learn • Laugh
Share • Celebrate



Jody and Trevor Strauss with Dr. Ghanashyam Acharya and his wife Sheetal Mhatre with Teresa Partington



Dr. Stéphanie Cherqui reviews her progress on stem cell and gene therapy



Juliebee and Dylan Craig share their hope for 5 year old Emily



Members of the adult panel provide insights on living with cystinosis



Dr. Jennifer Simpson presenting her research on corneal cystinosis



Dr. Paul Grimm shared research on kidney disease



Natalie Stack with twins Emma and Gracie Patterson



Rachel Young, Shamon Keizer and Jennifer Loglisci

underscores the importance of better drugs like Procysbi® to improve medical adherence.

We ended with another family discussion where people shared the challenges and silver linings of their journeys with cystinosis. There was a lot more happy crying and hope. We started talking about the *Power of Awesome*, when Erin Little surprised Nancy Stack with a beautiful quilt of the Day of Hope Tree, with pieces of fabric sent in by families from around the world. That was pretty cool.

A big highlight was seeing the new 2014 movie, which featured Hadley Alexander and her family. We were surprised and delighted to see clips of Sam and Lars in the video as well (even if it included a traumatic blood draw!)

We also heard from the Little family, who shared Olivia's story. The event raised \$2.3 million for cystinosis research, and thanks to the generosity of the Stack family,

every dollar will go to research.

It was hard to leave. That night Sam said, "I want to go to the babysitters tomorrow so I can play with my friends. I don't want to go home."

We spent the 11-hour drive home yesterday brainstorming on ways we can raise more money and help the cystinosis community. We are definitely counting down the days until next year.

Stephen Jenkins and his wife Ashton with their sons, Lars, age one, being held by his dad, and Sam, age four, at the 2014 Day of Hope Family Conference. Stephen was the unofficial journaler at the conference. The Jenkins family lives in Salt Lake City, Utah, where Stephen recently graduated from medical school.



Saturday night was the big Natalie's Wish event, which is always incredible. We got to present our check from Sam's Hope for a Cure, which included money raised in 2012 from our first letter campaign fundraiser, as well as the money raised in 2013 by Mary Ann Franson with her garage sale, totaling \$15,560.

We were impressed by how much money families like the Flerchingers, Sturgis, Cunninghams and Partingtons raised – over \$600,000 combined! During dinner we got to sit with the Smethhurst family from Logan, Utah and the Head family from Seattle.



Ghanashyam and His **Magical** Cysteamine Patch

By Stephen Jenkins, Samuel and Lars' dad, Salt Lake City, Utah

The most exciting surprise at the Day of Hope Family Conference was when Kevin Partington, Jenna and Patrick's dad, pulled up his sleeve to reveal a patch on his arm. He wasn't quitting smoking or going through menopause. The patch was a novel cysteamine delivery system developed by Dr. Ghanashyam Acharya.

Dr. Acharya was introduced to the cystinosis community at the 2013 conference, where he made quite a splash with his energy, enthusiasm and unbridled optimism. He was recruited to the fold by Dr. Jennifer Simpson to develop a better way to treat corneal cystinosis. Together they came up with a nanowafer that is placed on the surface of the eye like a contact lens.

The nanowafer releases cysteamine gradually for a day – and possibly up to a week at a time. Currently, the only treatment for corneal cystinosis is Cystaran™ eye drops, which must be applied every waking hour to eliminate corneal crystals.

Dr. Simpson has since used confocal microscopy and a mouse model of cystinosis to show that the wafer is much more effective at reducing corneal crystals than eye drops. Hopefully we'll have a clinical trial in the near future!

At the 2013 conference Dr. Acharya also talked about his experience using nanotechnology and microparticles to create long-acting drug formulations of felodipine, risperidone, progesterone and paclitaxel.

The new formulation of the antipsychotic risperidone can be given as an injection at six-week intervals, which has obvious benefits for people with schizophrenia and problems with medication compliance.

My mind started racing as I imagined what a long-acting version of cysteamine could mean for patients with cystinosis. We love Procybsi®, which is taken every 12 hours, but I still worry about the fluctuations in my son Sam's cystine levels. I was picturing something like a depot shot, given maybe once a week, or even once a day. That morning I talked with other families who had come to the same conclusion.

I was so excited about the idea that I submitted a question to Dr. Acharya, who was on the Q&A physician panel, to see whether he believed it was possible to create a depot shot for cysteamine. His response was simple and confident: "Yes, I can do that."

But there were naysayers on the panel, and their reasoning made sense. They said things like, "The amount of medication you need to deliver would make it impossible. We're talking about grams of cysteamine!"

A lot of people, including me, felt deflated by these remarks, but Ghanashyam insisted it could be done. The gauntlet had been thrown down, and the wheels in Ghanashyam's head started moving.

Fast forward one year to the 2014 Day of Hope Conference. Ghanashyam brought a prototype patch with him. After updating us on the cysteamine nanowafer for the

eyes, he gave us the quick run-down on how he developed the patch.

When he returned to Baylor after last year's conference, he rounded up his four post-docs, put their other research on hold and brainstormed for a solution. The main problem was the large amount of drug that needed to be delivered. Most skin patches, like fentanyl, estrogen and nicotine patches have very small amounts of drug (like micrograms), which allows the drug to passively diffuse through

mean stable cysteamine levels all the time, with better predictability of cystine levels in cells.

I talked with Ghanashyam at dinner on Friday night, and he told me that he loved to solve seemingly impossible problems. He said that you can never take no for an answer. He worked at NASA where he learned the motto, "failure is not an option." He also said that he is not in this business for the money. He just wants to find solutions,

silver linings of living with cystinosis, Ghanashyam's wife took the microphone for a moment. She said that right after they were married last year, Ghanashyam would often wake up at two o'clock in the morning and tell her he needed to go to the lab. She thought he was crazy, but after coming to the Day of Hope Conference and meeting the cystinosis families, she understands why he works tirelessly and with such urgency.

To much applause and laughter, she said she would never question his late night lab antics again.

Ashton and I joked at the conference that if we ever had another son, we were going to name him Ghanashyam. At the time it was an easy declaration to make, because after having a second child diagnosed with cystinosis, we felt like we were probably done having kids.

But with the patch and a possible cure in the pipeline, taking another roll at the dice might be in the future.

When asked whether it was possible to create a depot shot for cysteamine, Dr. Acharya responded with a simple and confident: "Yes, I can do that."

the skin and into the bloodstream. A cysteamine patch would require an active pumping mechanism, which has never been done before.

The physics were way over my head, but basically Ghanashyam invented a completely new model with multiple layers that are able to convert the mechanical energy of body movement into electrical energy that pumps the cysteamine into the skin.

He called it something like a "quantum kinetic transdermal patch." He thinks he can get 500 mg or half a gram of cysteamine into the patch, and his goal would be to have it last three days. The advantages of the patch are obvious. Medication compliance would go to 100 percent, and the peaks and troughs associated with every-six-hour dosing would be flattened into a continuous basal rate of drug delivery.

Hopefully this would mean less cysteamine would be needed per patient, and that side effects would be minimal. The patch might also solve the problem Dr. Dohil has pointed out: two hours after drug ingestion, cysteamine cannot be detected in tissue. The patch could

and to make life better for children and adults with cystinosis. I propose we clone one hundred more Ghanashyams!

On Saturday, when families were sharing the challenges and





Photo courtesy of Bob Hodson Photography

I HATE CYSTINOSIS

July 5th marks our three-year anniversary of life with cystinosis. It's a day we try very hard never to think about. After months of us telling our doctors something was not right with Olivia, our voice was finally heard.

After Olivia lied on the hospital bed almost dead with her kidneys shut down, we were rushed into a helicopter to get to the closest SickKids Hospital. One round of hemodialysis and a blood transfusion later and we were out of ICU. What a relief, we were moving to the pediatric floor. After a week and a half of blood tests, ultrasounds, x-rays and all the random poking and prodding, our medical team had narrowed it down.

Dr. Filer came into the room on Tuesday morning announcing that we would head over to see an ophthalmologist to rule out this rare genetic disease called "cystinsomething." Honestly we didn't even listen to him because, of course, there is no way our baby had a disease. Our ophthalmologist came in, introduced herself, took one look in Olivia's precious blue eyes, and without hesitation, the doctor announced, "She has crystals."

By Erin Little, Olivia's mom, Ontario, Canada, presented at the 2014 Natalie's Wish Celebration

When Nancy asked me to speak tonight, I was honored, scared and filled with joy to share our family's journey and give you a glimpse of what cystinosis brings to our family.

We dreamed of having a family and in 2009 that became a reality. I remember standing in the dining room and saying, "I'm pregnant."

Following the great news, I went out and purchased every "how to," "how not to do" and "what to expect" book. In all the books I read I never came across a section on how to prepare for a sick kid with a rare disease – and there was definitely not a section on "how to share your story."

We had no idea what this meant, but we were devastated. After three more stressful weeks of hospitalization, Olivia was finally strong enough to come home. We left the hospital with a laundry list of medications, supplements, bags of syringes and a binder full of information on how to ensure we are doing things right. Our life was turned upside down.

Today, life is pretty good and we live a pretty normal life – our normal is just a little different. Our house runs like most houses with a four-year-old and a 10-month-old. We mind our manners, play, get messy, have many time-outs, and we have a list of words we aren't supposed to say – and when I say "we," I mean "the kids."

Two words that come to mind are, "can't" and "hate." If Olivia hears you say these words, she will remind you that they are "bad words" and she will demand an apology for using them. Thankfully, Olivia is off enjoying a movie with the other children tonight, so I am going to use one of those bad words, but before I do that, I must tell you what a beautiful little girl Olivia is.

She is a strong-willed, busy, creative little princess who loves the color purple, coloring, playing in the garage and getting dirty. She loves books; she is the best big sister to

Harper; she gives the best hugs; oh, and, she has cystinosis. Most days are like everyone else's and "cystinosis" happens only four times a day: at 1 a.m., 7 a.m., 1 p.m., and 7 p.m. These are the awesome days.

The other day cystinosis seemed to happen all day long. It started at 6 a.m. It was wake-up time and as I approached Olivia's room I heard, "Mommy, my tummy hurts." I already know that breakfast is going to be a battle. Calorie intake is so important for Olivia. She needs to get so many calories a day to maintain adequate weight and height.

Meals aren't as bad as they use to be – Olivia has turned around 110 percent during mealtimes, and actually has a handful of foods she will eat. I am always her short-order chef. But, on this particular morning cystinosis won the battle.

I hate cystinosis.

Snack time rolls around and I know I'll win this round because Olivia is hungry, so calorie-packed muffins, here we go. Three-quarters of the way through snack, the front door opens and a polite good morning is spoken. It's LeAnn, the world's best nurse. She's here for Olivia's monthly Arnasep shot. It's a shot she receives to increase the production of red blood cells.

We are so grateful that a nurse comes to do this job for us. Tears begin to trickle down Olivia's face as she anticipates the pain. The brand new box of Mickey Mouse Band Aids don't even earn a smile. We pin her down; seconds feel like hours, finally it's over. Band aid applied, and an episode of Doc McStuffin makes the tears disappear.

I hate cystinosis.

Every Thursday, we have a playdate with a family friend – the girls run, play and make a mess. After two hours of intense play, I remind Olivia to drink some water because she has been running a mini-marathon around the house.

With the saddest face and softest voice she says, "Mom, if I drink water, I will have to go potty all the time and I will miss playing with my friends."

My heart breaks. **I hate cystinosis.**

Nap is followed by lunch. We read our nap-time story – today's choice, Giraffes Can't Dance. Olivia still has to wear diapers at nap-time due to the amount of water she consumes; nap-time water intake is generally 24 ounces. Olivia desperately wants to transition to wearing "big girl" undies at bedtime and every day when she wakes up soaked from the chest down, she says to me, "Maybe tomorrow I'll do better mommy." My heart breaks because I am so proud and sad. **I hate cystinosis.**

The remainder of this day unfolds nicely. All of Olivia's day care friends head home, dinner is served, bath time madness unveils, and she's tucked in by the best storyteller in the world, Dada. The words, "I love you," and, "See you in the morning," carry throughout the house.

Chad and I tend to our weekly preparation of Olivia's medication. We pull 308 syringes for the week. As we sit and share the happenings of our day, our conversation comes to a halt when we hear Olivia cry for us. When we get to her room, blood is pouring out of her nose. This is a side effect she gets from her monthly injection. The nosebleeds last anywhere from 10 to 20 minutes depending on the cooperation we get from Olivia.

**As we sit and share our day,
our conversation comes to a halt
when we hear Olivia cry. When we
get to her room, blood is pouring
out of her nose. This is a side effect
she gets from her injection.
I HATE CYSTINOSIS.**



Photo courtesy of Lars Wanberg



Tonight she was easygoing and it was over in 15 minutes. A quick change of linens and pajamas, and all is over. **I hate cystinosis.**

This is a day when cystinosis won.

Now I am going to contradict everything I just said but let me first explain. In our house instead of saying grace at mealtime, we all share at least three things we are grateful for that day. Most often, Olivia is grateful for her toys, and on extra special days – Mom, Dad and Harper are on her list.

Tonight, Chad and I are grateful for cystinosis. It has made us strong people, better parents, and we have met a lot of wonderful people along the way.

We are grateful for YOU, everyone sitting in this room.

YOU are making a difference for Olivia and every other cystinosis patient.

YOU give us hope and inspiration to continue to fight this battle.

YOU are here another year to help our children and for that we will be forever grateful.

And last, we are grateful for Nancy and Jeff Stack for their dedication to finding better treatments, and ultimately a cure. Cystinosis is not forever but the connections along the way are.

Thank you all for opening your hearts to our story. We will be forever grateful.



I am. Two of the most powerful words; for what you put after them shapes your reality.

-Author Unknown

My name is Olivia Ann Mae Little.
Running and playing is my game.
I wear pig tails in my hair and a smile on my face.

I am happy.

I love to dress up, putting mom's jewelry on
is my favorite thing.

I am beautiful.

I love to work in the garage with Dada.
He fixes the truck while I work on my coupe car.

I am handy.

I love to play outside and get dirty. All winter long I have
been playing in the snow pretending to climb mountains.

I am adventurous.

Sometimes I forget to listen and
I have to go on time out.

I am curious.

I love to read books and I
have a million questions.

I am inquisitive.

I have to take vitamins four times a day,
eye drops six times a day
and once a month I get a needle.

I am brave.

I love to paint and color and
I dream of unicorns and glitter.

I am creative.

Harper is my new little sister. I can make
her smile and laugh like nobody's business.

I am hilarious.


Sometimes I get sick, Mom and Dad say that's okay.
Now I know to go to the bathroom when I feel sick.

I am courageous.

I have no idea what cystinosis is.

I am innocent or I am four.

By Mom and Dad



Congratulations **GABE** *and Good Luck*

Our family has always thought of cystinosis as a stepping stone, not an anchor. Little did we know how literally Gabe would take this thought. He is currently making plans to attend college 12 hours from home.

Gabe is almost finished with his senior year of high school. We have been blessed to be able to homeschool him. We believe this has helped to keep him healthy and enabled him to do so well educationally.

In the beginning of our journey, Gabe was nauseated every morning to the point of not wanting to eat but still needing to take his dose. Homeschooling has allowed Gabe to get up, take his meds and then lie back down to rest for 30–45 minutes.

By the time he started kindergarten, he was on a schedule where he rarely threw up in the morning and he was able to focus more. During high school, he has had a very rigorous curriculum and still maintained a 3.9 GPA (4.0 weighted). He scored a 27 on the ACT and has an unbelievable memory. Now, we are continuing to be blessed as Gabe has been accepted to Pensacola Christian College, where he plans to major in mathematics.

We know research shows that many cystinotics struggle academically. When you consider that cystinotics on Cystagon® never get a good night's sleep and are extremely nauseated, how can they do well academically? They are tired and worried that they will need to run out of the classroom. But, we wonder, is it the cystinosis that causes learning problems or is it the lifestyle that goes along with having the disease?

It's been a long time since Gabe's diagnosis when he was only 13 months old. Those were the mornings of watching him projectile vomit. Those were the days of begging him to eat just a bite, the nights of changing sheets, and the alarms going off every six hours for the medication routine. Back then, it seemed he would never have a "normal" life, but we never gave up hope for his future.

As parents, we still have the typical anxiety of having a child on the verge of leaving home for college, but it is comforting to know that Gabe will be able to manage his medications. If he was still on the six-hour medication routine, he would probably live at home and commute to a local college. Procysbi® has become the next stepping-stone and it is allowing Gabe to follow his dreams.



Night of A Thousand Stars

At the 2014 Natalie's Wish Celebration on Saturday, April 5, 470 guests packed the Balboa Bay Resort ballroom in Newport Beach, California to capacity. That record-setting crowd, donated \$2.3 million for cystinosis research, bringing the total raised by the Cystinosis Research Foundation to \$25.8 million since 2003. The 2013 Natalie's Wish Celebration was the most successful fundraising event in Orange County last year, and our 2014 gala seems likely to be this year's top event.

It's impossible to talk about the annual Natalie's Wish Celebration without mentioning the attendees themselves. The room is always filled with caring guests, many first-time attendees, who have some understanding of what to expect from the evening – but they often leave the event totally moved, and at times, teary-eyed, by what they have just experienced.



On April 5, guests were dazzled by The Tenors, four talented young men who left the audience begging for more. After a sumptuous meal, guests watched an emotional video filled with the hope, courage and commitment for which cystinosis patients and their families have become well-known; and, as usual, guests were blown away by an over-the-top live auction.

But ultimately, attendees were mesmerized as two mothers shared heartbreaking stories about their cystinotic children. It is easy to

understand Nancy Stack's relentless passion and why Erin Little "hates" cystinosis. But it is also easy to see why Nancy and Erin remain so hopeful since each Natalie's Wish Celebration brings us closer to a cure for cystinosis.

We still have far to go on our journey to find a cure but the generosity of Natalie's Wish attendees continues to give real hope to the children and young adults in the cystinosis community.

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



Sandy Sewell with Brenda and Tom Melang and their daughter Sarah



Tom and Traci Gendron with Nancy and Jeff Stack



Shelley Belling, Shannon Tarnutzer and Karen Linden



Natalie and Tom Moran, Jr., with Susan and Tom Moran, Sr.



Christyne and Bob Olson



Cindie and Bert Selva



Spirited bidding by Peter Rooney during the live auction



Lora Carpenter, Natalie Stack and Pooja Sharma

Little Fighter with a Smile

By Nicole Manz,
Keegan's mom,
Morrisville,
North Carolina



*Keegan Manz; his dad, Brad;
his mom, Nicole; and brother, Shane*

Keegan will be a year old on April 23, 2014. He was a perfectly healthy, 22-pound baby boy until age five months. Then something happened and he was not right. His eating pattern changed. I was told this was normal; babies start to stretch out their feedings as they get older and he was just taking more each time he nursed.

But other odd things began to happen. His bowel movements dropped to once a week; he started to get carsick; gag when he nursed; and lost four pounds in just a couple weeks. Our once-happy, smiley baby was so lethargic that he did not even want to be held or touched. All these symptoms were brushed off by a pediatrician. After four doctor's visits, I demanded to see our regular pediatrician. One look at Keegan, and he immediately sent us to the ER.

Something was really wrong. As I loaded him into his car seat, I promised him that we would get through this together and I would get answers to make him better. At the ER he had a battery of tests. When his potassium came back below 2.5, they admitted him for more tests. Within a couple of hours we were rushed to the PICU. Keegan's potassium had dropped so low the doctors were worried that he would go into cardiac arrest. We were told that had we not been in the hospital, he probably would not have lived through the night.

Little did Brad and I know this was the beginning of our journey with cystinosis.

The next three weeks were every parent's worst nightmare. Day after day more tests were being done – yet no answers. I cried and cried while spending hours on the computer trying to figure out what was happening to our baby. Finally, he was diagnosed with Fanconi syndrome. The doctors started to prepare us for

Keegan is so resilient and he brings so much light into our world. He has what it takes to fight this battle with a smile.



some pretty scary stuff. They had ruled out cystinosis because his calcium was so high, and he had been in the 100 percentile in height and weight since he was a month old.



However, as I searched the web I came across many stories so similar to his that I knew Keegan had cystinosis.

On November 13, 2013, three weeks after our trip to the ER, the phone call came: it was cystinosis. I spent the next few days crying as Keegan's future seemed very dark. It took a few days but I finally realized that I was not doing anyone any good and pulled myself together. Brad and I promised ourselves that we would make the best of this. We would find our "new normal."

To be honest, the next few months were very rough. Keegan was sent home with an NG tube and a strict feeding schedule. Our days were spent trying to keep him from throwing up. Keegan did not touch the floor for almost two months. He was very sick from his high-volume feeds and barrage of new medications. It took us about eight weeks to work up to the full dose of Cystagon®. With each increase Keegan's little body would fight the medication.

Our days were pretty much spent changing clothes and holding our sweet baby. "This too shall pass," was my mantra for those eight weeks. By mid-January, he was up to full

dose for his weight and we were finding our norm. We were fortunate to have family who came to help us for the first couple of months.

What has amazed me the most is Keegan's spirit through everything. He is such a happy baby. It is not uncommon for him to throw up – and then moments later be laughing and smiling. He is a blessing and he has taught us so much. He keeps us smiling through the tougher moments. Everyone at Duke has fallen in love with Keegan. Even the lab technicians are amazed that he rarely cries during blood draws or finger pricks. He is so resilient and he brings so much light into our world. I know he has what it takes to fight this battle with a smile.

We have been blessed to have a great support system. Our family, friends, neighbors and strangers have gone above and beyond to show their love and support.

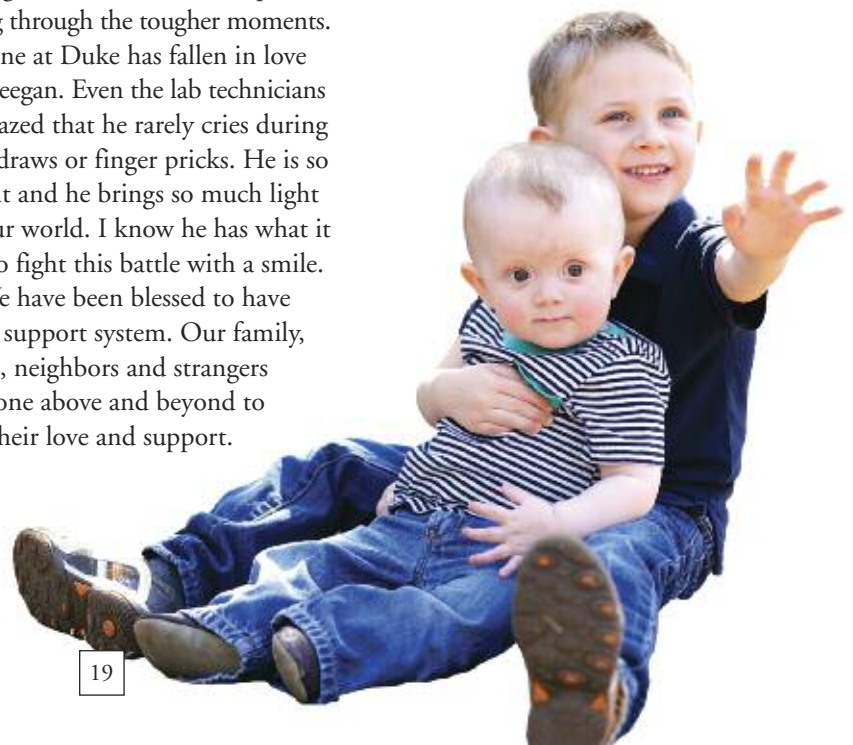
We've had some challenging moments but through the dark, the light has shined and we thank everyone who has supported us so far.

Facebook has been God's blessing for me. I have connected with so many people who are walking this path. I truly feel like I have a second family. We were sad not to be able to attend this year's Day of Hope Conference, but we are excited to meet everyone and get involved. Rain or shine we will be there next year.

Despite the challenges we faced the first few months, and every day, Brad and I have a positive attitude. We know that Keegan will be okay. He will have a life. He will know love. He will be whoever he wants to be.

I would not wish cystinosis on anyone. However, I do realize that having cystinosis is not the worst thing either. We almost lost our baby. A life with cystinosis is better than no life at all. I am grateful that we live at a time when a treatment is available, and when so many medical breakthroughs are so close at hand.

Finally, we are fortunate to have Nancy and Jeff Stack, and so many others who support the Cystinosis Research Foundation. Working together we will find a cure.



What is a BFF?



Christine and Duane Eddy

A Best Friend Forever is someone who faithfully stands beside you no matter what. A BFF is with you during the storms of life, they provide a shoulder to cry on and extend a hand to lift you up. They go out of their way to help and give selflessly.



*BFFs Amanda and Elizabeth
Anne Kuepfer and Chloe
and Gabbie Strauss*

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

I am blessed with such a BFF. Her name is Christine Eddy. Our friendship started 28 years ago when we were in fourth grade. We went through elementary school, high school and university together. We saw each other get married, buy homes and have babies. Christine had two beautiful healthy baby boys, Chase and Ryder. I had two beautiful baby girls, Gabbie and Chloe, but Gabbie was not born perfectly healthy. Gabbie was diagnosed with cystinosis.

I remember sitting in a waiting room of the pediatric ward at Grand River Hospital. My mom was there to relieve me from the exhaustion of being seven months pregnant and having a 15-month-old admitted, hooked up to IV and receiving countless blood draws. I took a break and called my BFF. I cried and she listened. Christine has always asked about Gabbie's health. I knew that she really cared.

So, what does a BFF do when their best friend's daughter has an orphan disease and needs research money to find a cure? They rise to the challenge, they sacrifice time with their family, and they give selflessly. Christine, a busy full-time working mom of two young boys planned a fundraising dance. Her husband, Duane Eddy, lead vocalist and guitar in the Sawmill Road band offered his time and talent. He and the other Sawmill Road members – Rob Hood, Noel Savage and Shane Chapman – volunteered to play their heart out for Gabbie without asking anything in return.

While in the midst of planning, Christine phoned to share that she was contacted by a lady who had a child with cystinosis. I thought, 'really? Could there be someone from this mostly rural area affected by cystinosis'? It seemed unbelievable. Sure enough, I had the privilege of meeting Julia Wells, her husband Michael and daughter, Rachel that night at the dance. Julia had lost her son, David, to cystinosis when he was just five years old in the 1970's. They lived in England at the time. The pain of their loss hit me. Gabbie is six. I'm sure Julia would have loved another year with David. Julia's story reminded me that each day and each moment with our children is a gift.

I was amazed by the strength of Julia and her family. The pain they experienced as a result of cystinosis did not stop them from giving their love and heartfelt support. They too have hope and believe this disease will be cured. Surprisingly, I was the first cystinosis mom Julia had ever met. I couldn't believe it! Look at how far we have come in offering support to parents, friends and family affected by this disease. Look at how our cystinosis community has united. Can you imagine being Julia? Can you imagine having no one else to talk to? Julia reminded me that I have much to be thankful for. As difficult as some days can be, nothing can be more difficult than what Julia has endured.

My BFF's hard work and effort came together on November 9, 2013. The Legion in the village of Colborne with a population of just over 2,000 was packed with people supporting cystinosis research. The \$10 tickets were sold out. Sawmill Road was amazing and the dance



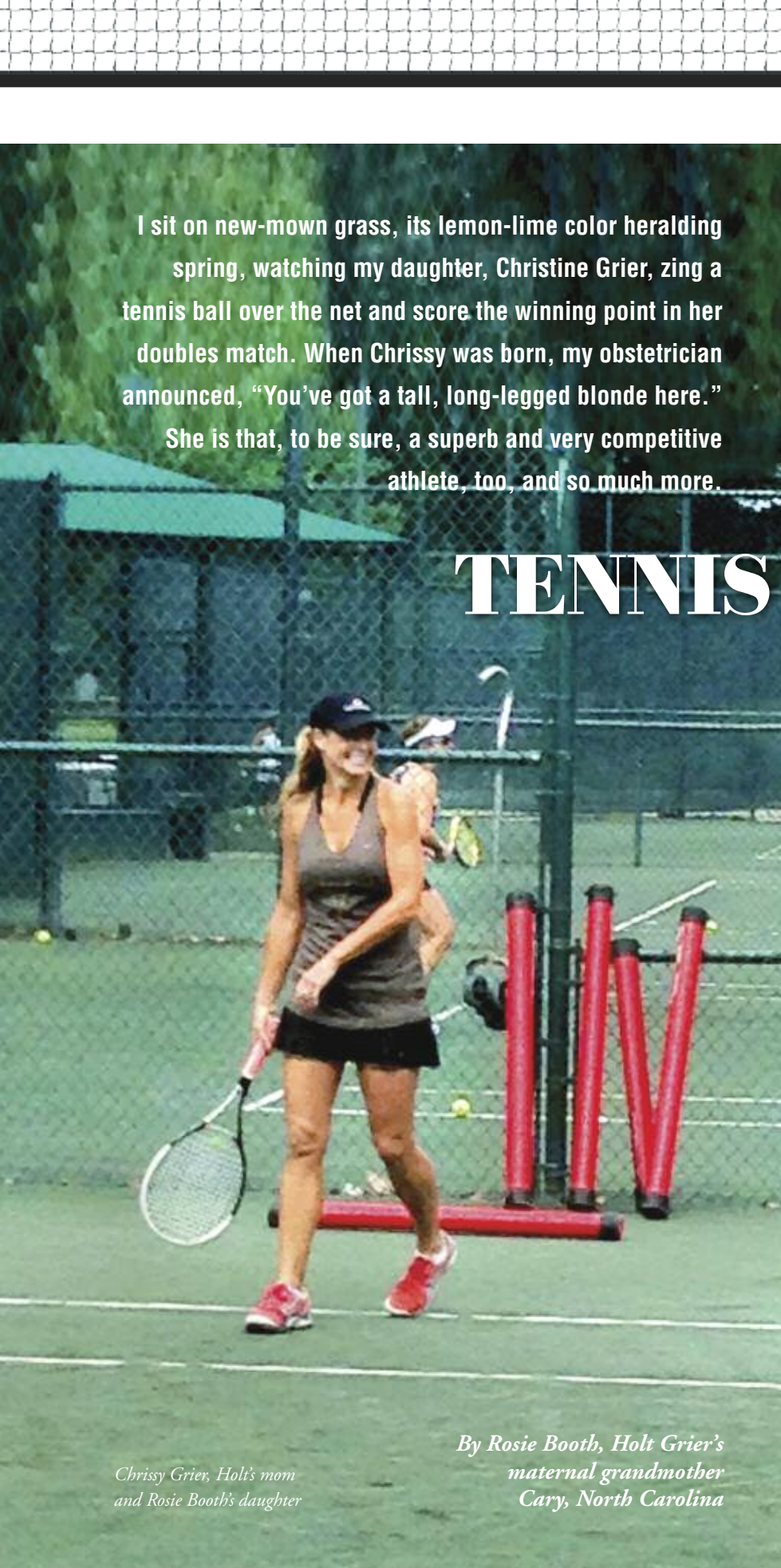
Chloe and Gabbie Strass at the 2014 CRF Day of Hope Family Conference

floor was full. The silent auction item table was busy all night, as were the volunteers selling raffle tickets. Every heart in that room was generous. Colborne may be small, but the community has a big heart. Over \$7,000 was raised that night! I have the best BFF in the world!

A few months after the dance, another small town with a big heart raised money for cystinosis research. The Paradise and District Lions Club of St. Clements, Ontario held their 6th annual Tree of Lights campaign. The campaign was their most successful effort yet, raising \$2,639 for research! We are so thankful for the love and support of the St. Clements community. Every dollar raised is a dollar that makes a difference.

I am so grateful for each friend, family member and community that has united to help raise money for the Cystinosis Research Foundation. Thank you for sacrificing your time, your talents and your money to make a difference for Gabbie and all the families affected by cystinosis. Our family continues to hope and pray that a cure will be found soon.

A BFF is with you during the storms of life, they provide a shoulder to cry on and extend a hand to lift you up. They go out of their way to help and give selflessly.

A woman with blonde hair, wearing a dark grey tank top, black shorts, and a black cap, is playing tennis on a green court. She is holding a tennis racket and looking towards the camera with a smile. In the background, there is a chain-link fence and some red equipment.

I sit on new-mown grass, its lemon-lime color heralding spring, watching my daughter, Christine Grier, zing a tennis ball over the net and score the winning point in her doubles match. When Chrissy was born, my obstetrician announced, “You’ve got a tall, long-legged blonde here.” She is that, to be sure, a superb and very competitive athlete, too, and so much more.

TENNIS ANYONE?

A psychology major in college, she used her training to listen to people’s needs and desires as she “counseled” them as they shopped for a new home. Her career in real estate, which she considered a sort of ministry – trying to find just the right home for each client – was blossoming.

Then came Mary Logan, Jack and Holt. Chrissy and her husband, Jason, embraced family life. Jason’s business success accelerated and Chrissy assumed the unsung-hero role of stay-at-home mom.

The children seemed to be thriving, even though Chrissy’s third pregnancy was deemed high risk. After the birth of her second child, Chrissy had developed something called Antibody E, discovered when she donated blood. If during pregnancy this antibody crosses the placenta, it can severely damage the developing baby. While pregnant with Holt, Chrissy was closely monitored, and nearly three weeks before term, lab results suggested that labor be induced. All of Chrissy and Jason’s large extended family rejoiced when Holt was born healthy, and mother and baby – and also Dad! – were doing fine.

Flash forward 14 months. It’s a Friday night in February and I am hosting a birthday dinner at home for one of Chrissy’s four sisters. The phone rings.

“Mom,” Chrissy begins. She is weeping. “Jason and I are on our way to the ER with Holt.” I knew that Holt had been sick for several days. But it was winter. Holt’s siblings had been fighting a virus. Holt had been seen by his pediatrician earlier in the week. That Friday morning, however, Chrissy had called me to say she’d insisted that he be seen again that afternoon. Tests had been run and Chrissy had taken Holt home and put him to bed for the night.

*Chrissy Grier, Holt’s mom
and Rosie Booth’s daughter*

*By Rosie Booth, Holt Grier’s
maternal grandmother
Cary, North Carolina*

At 7 p.m. the doctor's office called Chrissy with the test results. "Mrs. Grier, wake Holt immediately and take him to the ER. His potassium levels are so low, his heart is in danger of failing."

"So, can you come to us right away, Mom? Chad (Jason's brother) is with Mary Logan and Jack until you and Pop can get here."

Within 15 minutes my husband and I began the three-hour car ride from our home in Cary, North Carolina, to Holt's home in Charlotte. And so began our introduction to cystinosis seven years ago. We quickly learned that this devastating disease is caused by a defective recessive gene in each parent, and according to the laws of genetics, each pregnancy has a one in four chance of resulting in a baby stricken with cystinosis. Chrissy and Jason and all of us had never heard of this illness and had never known anyone in our families so afflicted.

So how could this have happened and why?

Early on Chrissy said, "I have to believe that there must be some purpose in this, some reason this has happened to our little boy and our family, and maybe that purpose is that we can do something about it."

Chrissy and Jason quickly found information online and discovered the Cystinosis Research Foundation (CRF), and an entire support network led by Nancy and Jeff Stack. In addition, Holt's pediatric nephrologist, the dedicated and knowledgeable Dr. Charles McKay, who in his long career had treated children with this rare disease, provided guidance and calm reassurance.

"Holt will grow up," he said, "and since 1994 we've had a treatment that allows kids to reach adulthood, whereas prior to that they died between six and 12 years old."

The hope that a cure will be found in Holt's lifetime sustains us. Indeed, we are heartened by the

development of new medicines and treatments that are leading to improved quality-of-life. The Griers and the extended Grier clan, with special thanks to Holt's aunt, Aubrey Grier, host an annual gala sponsored by the Hope for Holt Foundation that has raised over a million dollars during the past seven years.

tennis match, I wonder if Holt will one day have the stamina to race all over the court and slam the ball over the net. The good news is he's already taking tennis lessons and this past fall he graduated from half-court to full-court basketball. He's a great reader, an accomplished writer and illustrator, sings in his school's Show Choir, and



Holt Grier with his grandmother Rosie Booth

Friends and neighbors have organized a walkathon, a golf tournament, a Corn Hole Challenge, the sale of handmade angel earrings, and arranged for an annual donation from an adult Sunday School class. Also, Jason for the last several years has served on the board of the CRF.

Meanwhile, on this grassy slope as I applaud my daughter's successful

has a great sense of humor. I can't wait to see what he'll become when he grows up.

Right now, for Holt with no cure yet available, it appears the score is Advantage Out, but with the love, support and prayers of his mom and dad and his big extended family and the whole CRF community, he WILL grow up and he'll win this match!

7TH ANNUAL

Hope for Holt Gala



The 7th Annual Hope for Holt Gala, held on Saturday, January 25, 2014, at the beautiful Charlotte Country Club was a record-breaking success.

This highly anticipated event, held in honor of Holt Grier, featured spirited silent and live auctions, a delicious dinner, dancing far into the night – and is the year’s most fun-filled party. Best of all, it raised \$220,000 for cystinosis research.

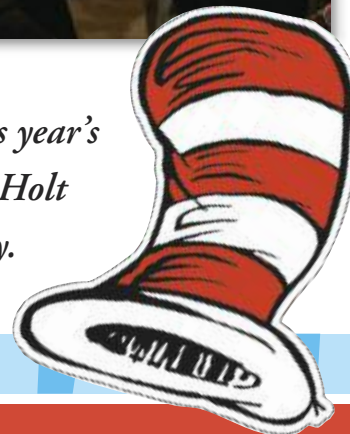


Congratulations to Chrissy and Jason Grier, Hope for Holt Foundation board members, and the many dedicated volunteers who work tirelessly throughout the year to make Holt’s gala so successful.

Proceeds from this annual sellout are sent to Hope for Holt Foundation’s partner, the Cystinosis Research Foundation, who uses every dollar it receives to fund research to find a cure for cystinosis.



Thank you to everyone who attended this year’s gala. Your support is giving new hope to Holt and everyone in the cystinosis community.



We’re already planning next year’s event. For details visit hopeforholt.org



Faith Hope & a Cure

I dread when I'm asked to write an article. I do not like to go to that place where reality sets in. Most days I can see the silver lining of cystinosis, while on other days I cry and wonder why things have to be difficult for Tina. Mark and I full-heartedly believe there will be a cure for her, however it cannot come quick enough. Our entire family is exhausted, but we will continue to forge ahead.

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

This year's Day of Hope Family Conference was incredibly inspiring. It started as each family shared their greatest wishes for their children. The words "cure" and "normal" were used frequently. Jean Blum shared that her four-year-old son, Jackson, wished that "his friend Tina" would be cured. Ironically, I'm sure that he doesn't realize that he too has cystinosis.

Tina has experienced the disappointment of missing several sleepovers during the past few months. So when she came home this week, teary-eyed because her class is planning a campout, and she knew it would be impossible for her to go, Mark quickly announced, "You ARE going, and I'm going with you." Needless to say, she is over the top about her first overnighter.

Recently, Tina received a Godly character award for endurance and persistence. Persistence is defined as: perseverance in spite of all obstacles and discouragements. This describes Tina perfectly. As Tina was handed her award, her teacher, Mrs. Downs, stated, "Tina is a strong soul. She has had plenty of obstacles yet maintains a happy, wonderful spirit. She is constantly thanking God for the wonderful blessings in her life." Indeed, there is much I can learn from this little Angel whom God has placed in our lives.

*Tina just wants
to be a typical
10-year-old.*

The other day Tina approached me with the dreaded statement, "Mom, I wonder what it is like not to take medicine." As my heart sank, I reassured her that she will soon know. On one occasion she asked why she must take her medicine if she is going to be cured. I spent the next 30 minutes explaining the job of each of the 19 pills she would take that morning and the impact that cystinosis has on her failing kidneys. I then sent her off to school, as I barely managed to hold back tears. That's a lot for a 10-year-old child to take on, and I know that Tina, being the awfully intelligent child that she is, will spend her time pondering what I have just told her. I dread the day she reads or hears what cystinosis really is. I'm unsure how long we can shelter her from the realities of this fatal disease.

We were recently told that Tina only has 50 percent kidney function. We know a kidney transplant is in her future. We continue to pray that God will keep her strong until a cure is found. But we have great hope – research is being done every minute of every day by scientists around the world.

We have faith that a cure will soon be found for Tina, Jackson, and everyone Tina holds dear. Faith is believing in what you cannot see. Although this can be difficult in the midst of a storm, I must remind myself it gets hardest toward the end of the race. However, the finish line is in our sights, we are so incredibly close!

We are eternally grateful to everyone who has embraced Tina and walked this journey with us – together we are united in a cure!



NEVER DOUBT THAT A SMALL GROUP OF COMMITTED CITIZENS CAN CHANGE THE WORLD; INDEED, IT IS THE ONLY THING THAT EVER HAS. MARGARET MEAD

Small Town,

Big Heart



Tina having fun drawing raffle winners.

ten-year-old Tina Flerchinger, who was diagnosed with cystinosis when she was 17 months old, lives in Clarkston, Washington, with her parents, Denice and Mark Flerchinger, and her sisters, 19-year-old Nichole and 18-year-old Catherine.

Clarkston, near the Washington/Idaho border, is located in the Lewiston Clarkston Valley, which has a population of 61,300 – a relatively small number.

And yet, guests who attended the *Sixth Annual Wine, Stein and Dine* event on Saturday, May 17, had irrefutable evidence that residents of Lewiston Clarkston Valley have very big hearts.

A dedicated group of Tina's friends worked for many months planning the event, which is now the most anticipated in the community. Their hard work and attention to detail was evident throughout the evening. Even the local meteorologist chipped in, providing perfect weather.

It was by every measure an incredible evening – one that raised \$110,000 for cystinosis research. All costs are underwritten, so one hundred percent of every dollar raised goes directly into the hands of cystinosis researchers who are trying to find a cure for the rare, genetic disease that destroys all of the body's organs and causes great pain in the process.

The Flerchinger family is enormously thankful for the extraordinary generosity of Rogers Toyota Scion, who again hosted the event in its beautiful showroom; to Basalt Cellars Winery and Riverport Brewing Company; Sodexo Catering; Duet Riendeau; and everyone who donated one of the 145 items that were part of the lively auction.





Mark and his mother, Barbara Flerchinger



Tina with longtime supporter Jack Seeh



Tina's Hope board members (standing) Sherry Seeh and Mary Ausman, applauding the spectacular Fund-a-Cure

The event had an international flair, as cuisine from Paraguay, Nigeria, Vietnam, Native America, and Russia was served.

But when all was said and done, this was an event about a caring community in Washington and the surrounding area, that came together yet again to help Tina and others with cystinosis. Their generosity is already providing an improved quality-of-life for those around the world with this dreaded disease, and real hope for a world free from cystinosis.



Generous donors Arnold and Carol Beckman with Mark and Denice Flerchinger



Tina with her sisters, Nichole (l), and Catherine (r)



My Journey with Seth



My name is Leif and I'd like to tell you about my favorite person in the world, my brother Seth. Seth and I are fraternal twins and we began our journey together in the outside world on April 17, 2012. Though I was born two minutes earlier, Seth was heavier, and at 6 lbs., 14 oz., weighed 4 oz. more than me.

Seth is a beautiful human being inside and out. With blond, curly hair and bright blue eyes, Mom and Dad sometimes say that he looks like a real surfer dude. He is a happy, kind, gentle spirit, and is someone I can always count on for a giggle or a hug when I feel sad.

Seth is also a wonderful friend and companion. He has a great sense of humor and can always make me laugh, especially when he tells me jokes in our own special language, which Mom and Dad can't understand. Our favorite pastimes include chasing squirrels, watching birds, eating dirt and tomatoes from our garden, going for runs, and bike rides with Mom and Dad, stacking blocks, taking things out of the kitchen cupboards, and blowing bubbles at bath time. We also love dancing, and while I love to tap, Seth is more partial to whirling like a Dervish, spinning round and round in time to music composed by our favorites – Bach, Mozart and Raffi.

Seth started crawling a couple of months before me and it inspired me to get moving. I started walking just after our first birthday, but Seth took two months longer. Even after he had been walking for a while, we noticed that he was unsteady and fell over a lot. This seemed to make Seth really sad, and though I tried to cheer him up by sharing my sippy cup or soother with him, it didn't always work.

At about the same time, Mom and Dad started to notice a few other things about Seth that caused them to worry. Seth's growth had slowed down significantly compared to mine. He was also very thirsty, and Mom told people that he was sometimes inconsolable at bedtime.

didn't exactly understand what that meant, but I remember that sometimes it was hard for me to get to sleep at night because Seth was crying so much.

When Seth went to see an endocrinologist, she said that there might be something wrong with his bones. When she used words like "rickets" and "failure to thrive," I noticed that Mom and Dad looked really scared and I couldn't help but feel the same. What would follow was a whirlwind of appointments and blood tests. Though Seth was getting lots of special attention and colorful Band-Aids on his arm, I knew that he wasn't really having all that much fun.

On October 21, just after Seth and I turned 18 months old, we got a phone call. Dad left the room to talk. When he came back, he had tears in his eyes and hugged all of us close together. That was when our family found out that Seth has a condition called cystinosis.

Though I don't have cystinosis, it has become a big part of my life, of our lives. We spend a lot of time at the Alberta Children's Hospital with nice people like Seth's nephrologist, Dr. Midgley. He is one of my favorites not only because he is a twin, but also because he is funny and treats all of us with kindness and compassion.

Seth has to take five kinds of medicine every six hours day and night for the rest of his life, and Mom and Dad say that sometimes they feel like pharmacists in training. I often hear Mom and Dad sneaking into our room late at night to give Seth his medication, and I say a special prayer that it doesn't make his tummy hurt and that it will help keep him healthy and strong.

Cystinosis is scary. Though Mom tries to hide it, she sometimes cries

when she hugs us and I can tell that Dad is trying extra hard to be strong. It isn't easy. I have heard Mom and Dad talking about how uncertain the future sometimes feels and about their fears about what it could hold for Seth, for all of us.

perfect name for my brother, because he plants seeds of goodness, touching people with his joyous, gentle nature wherever he goes.

Seth is my hero and my best friend. I want him to live a long, happy, healthy life. I want our



But we are so thankful that Seth is doing well right now. He is so brave and takes his medicine without even the tiniest complaint. He is growing well, laughing, playing and just really loving life and everyone around him. One meaning of the name Seth is "to plant or sow." I think this is the

journey together to be long and full of adventures like camping, fishing, hiking, skiing, traveling and gathering tomatoes and raspberries from our garden. I want to explore the world with Seth by my side.

p.s. There's a note from my parents on the next page.

By Kristen Murray and Nathan deBruyn
Seth and Leif's mom and dad
Calgary, Canada

Coming to terms with Seth's diagnosis and our new reality has indeed been a challenge. When we learned that Seth had cystinosis, we were shocked that he had such a rare and serious disorder and devastated that our beloved son may suffer as a result of it. Awash with feelings of helplessness, fear and anger, we were dismayed that cystinosis had so insidiously entered our lives – an uninvited and unwelcome intruder that would force us to deviate from the journey we had envisioned for our family and for our son.



Photo courtesy of
Lars Wanberg

Heroes In Our Midst

However, during the first five months on our new path, we have been surprised and uplifted by heroes along the way. The first of these heroes has been the nephrology team at the Alberta Children's Hospital and the unfailing support they have provided.

This team, lead by pediatric nephrologist Julian Midgley, has treated us with kindness, compassion and unequivocal professionalism at each turn of our journey, guiding us through the administration of Cystagon® and the complex array of supplements that Seth requires and carefully monitoring his well-being. Providing awe-inspiring care, this team has embraced us as valued counterparts, inspiring and empowering us to educate ourselves about cystinosis and to play an active, even leading role in Seth's treatment.

After Seth's diagnosis, we soon felt the need to share with our loved ones and emailed our story as told by Leif to our circle of family and friends. Mere moments after we pressed the 'send' button, emails began to pour in expressing a range of thoughts and emotions that we hadn't anticipated. Though many people shared sadness and shock at our news, many more shared their optimism and hope. Many shared personal stories of challenge and loss and strength in the face of adversity. Everyone expressed love, compassion, empathy and heartfelt desire to help. And so, heroes have emerged from our midst.

We have been deeply moved by such outpouring of support and are buoyed by the spirit that surrounds us. We have been touched by the steady

stream of food, gifts and time that people have so generously shared. We have also been astounded by the many donations given to the Cystinosis Research Foundation in Seth's honour – more than \$32,000 to date.

Every dollar donated to the CRF is used to fund a plethora of exciting and promising research. We came to realize the power of the CRF at the recent Day of Hope Conference in Newport Beach, California, where we became more familiar with the incredible advancements that the foundation is making toward better treatments and a cure for cystinosis. We heard research updates presented by CRF researchers – some of the brightest scientific minds in the world – and we were heartened by the dedication with which they are working.

Laser-focused and committed to our cause, researchers such as Dr. Stéphanie Cherqui, Dr. Ghanashyam Acharya and Dr. Jennifer Simpson are developing nanotechnology corneal drug-delivery wafers, time-release cysteamine patches and autologous stem-cell therapy techniques for cell

As we heard stories of diagnoses, medication regimes, hospitalizations, polyuria, G-tubes, corneal crystals, muscle-wasting and kidney transplants, we were reminded of the havoc that cystinosis can wreak.

But we were also inspired by the everyday heroism of the parents,

bravely faced frequent blood draws, countless doctor's appointments and x-rays. Even during a recent hospital stay that was required when he caught a bug and was unable to tolerate his medication, Seth took the painful insertions of NG tubes and IV lines in stride, casually swinging his tubes over his shoulder so they would not impede his playtime. Cooped up in a hospital room for a week, Seth made the best of the situation, finding fun in reading books, playing with stickers, laughing with visitors, and despite his fatigue, always mustering energy to greet his doctors with a smile and a hug.

Seth embodies life and love and happiness, and his courage in the face of adversity has inspired us to stay strong. His presence has reminded us of how fragile and precious life is and of the importance of living each day with positivity and presence. And for that, we are truly grateful.

Our journey with cystinosis is certainly not one that we would have chosen. When Seth was diagnosed, we thought that it was the worst thing that could happen to us and that we faced a journey that could only be dismal and terrifying. But as we have crossed paths with so many remarkable and fearless heroes, we see that cystinosis is not the worst thing that could happen. No, the worst thing that could happen would be to overlook the passion, dedication, friendship, hope, love and joy that surrounds us as we navigate life with cystinosis; to forget that we are in so many ways really and truly blessed.

The worst thing that could happen would be to overlook the passion, dedication, friendship, hope, love and joy that surrounds us as we navigate life with cystinosis; to forget that we are in so many ways really and truly blessed.

repair. Fiercely dedicated to their work and to our children, sacrificing time with their own families, these brilliant scientists are working around the clock to make a cure for cystinosis a reality in the near future.

The Day of Hope Conference also allowed our family to meet the indefatigable Nancy and Jeff Stack, the foundational and driving force behind the CRF, and to witness firsthand their passion, determination and generosity. Brave leaders and staunch advocates, Nancy and Jeff are beacons of light on pathways that for so many would otherwise have been hopelessly dark. As they humbly and fiercely work, Nancy and Jeff leave no stone unturned in their quest to find a cure. Inspiring and empowering us to join their efforts to save our children, Nancy and Jeff are heroes who offer us a reason to believe that a cure for cystinosis is on the horizon.

In sharing our stories with other families affected by cystinosis at the Day of Hope Conference, we quickly connected with a group that embraced us as family, feeling kindred through a rare, devastating disease that 'outsiders' could never really understand.

grandparents, extended family who exude courage and strength in the face of this devastating disease, graciously accepting challenges and doing everything in their power to provide their children with the best possible care.

With unconditional love and purpose, these people never give up, awaking each day with renewed commitment, setting aside their own fears, exhaustion and frustration to make life brighter for their children.

With so many heroes in our midst, we cannot forget the most heroic of all: the children living with cystinosis and our beloved son Seth. We have been inspired by Seth's gracious and fearless acceptance of a relentless medication schedule. Taking 18 doses of medication each day, Seth follows a rigorous six-hour schedule, being wakened in the middle of the night with rarely a hint of complaint. He has





Swing and Bling #4

View Carre'



The Fourth Annual Swing & Bling Event to benefit Jenna & Patrick's Foundation of Hope took place on February 21 and 22 and was a great success – raising over \$235,000 for cystinosis research. As always, these funds will be passed along to the Cystinosis Research Foundation, where they will be sent to researchers who are doing the critical work of finding improved treatments for cystinosis. We are grateful to our friends and family who once again helped make the golf and dinner events such a success. So many people contribute in so many ways; when it all comes together you can't help but feel the good "vibe" that surrounds each facet of the event.



*Nine-year-old Patrick Partington
with his very proud mother
Teresa Partington*



*Nine-year-old
Jenna Partington*



Photos courtesy of April Melarkey Photography



The Stack Family

On a late afternoon in March 2006, after several months of visiting doctors and trying to learn what was wrong with our twin babies; and just weeks after Jenna had come home from a 13-day stay at Sutter General Hospital (half that time in Pediatric ICU); we received the call that our kids had a rare disease called cystinosis. It was simply incomprehensible. We had no idea what to do next.

That evening, I received a phone call from Nancy Stack who took the time to hear our story and share hers as the mother of a child with cystinosis. She and her husband, Jeff, would become our mentors and our friends, as they lovingly helped us make sense of how to deal with the devastating diagnosis.

Their daughter, Natalie, would become an inspiration and a comforting vision of hope. She is a special person for our children to look up to – we admire her great strength and willingness to do what it takes (and it takes a lot) to stay as healthy as possible. Natalie just completed her undergraduate degree at Georgetown University; and will begin work on a Masters Degree in social work at USC. We are happy to have her back home on the West Coast, and we are honored to have her here with us tonight.

February 22, 2014



Dear Family and Friends,

Thank you for taking the time, for making the generous investment and for showing your love by being at this event tonight. We are so grateful to this community for the long-standing commitment you've made to support cystinosis research. Kevin and I pray that one day these events may become obsolete, having turned all the prayers, money, and effort into a cure for Jenna, Patrick, and many others with the genetic disease cystinosis, and related illnesses.

There are several young people present tonight who share a big part of Jenna's and Patrick's story. Natalie (22 years old), Joe (17 years old), and Hadley (4 years old) are all present in this room, they all have cystinosis, and they are all taking the newly FDA-approved drug, Procybsi® every 12 hours, every day; as they do their best to keep their progressive disease at bay. They are all very brave, tough, scared, mad, grateful and ultimately very human. I hope you will take the opportunity to meet them and their families this evening.

The Bling event is a celebration, and it's fun! All the same, it's a celebration inspired by our children, who have a progressive, incurable disease. Sometimes it's tough to think about putting on a party dress when I think too much about what the party is for. However, we feel upbeat and excited to celebrate when we think of the spirit of community, the friendship and love that have become such a big part of this event.

When I consider it carefully, I realize Jenna and Patrick aren't inspirational because they have a disease; they are inspirational because they have taught us all so much. For that, we celebrate!

Love, Teresa and Kevin



Heartfelt Thanks

Through your kindness and generosity, you have given hope to those with cystinosis and brightened their days.

There are no greater gifts.

In 2013, donors from around the world helped CRF raise \$3.2 million for cystinosis research.



Jenna & Patrick Partington
\$251,225



Holt Grier
\$167,555



Henry Sturgis
\$130,100



Tina Flerchinger
\$119,005



Landon Hartz
\$31,536



Joshua Clarke
\$21,040



Bailey DeDio
\$17,662



Gabbie Strauss
\$16,743



Andrew Cunningham
\$10,048



Sam and Lars Jenkins
\$9,656



Nicole Hall
\$8,183



Olivia Little
\$4,810



Jake Krahe
\$4,680



Mary Head
\$4,523



Camden Sanders
\$4,500

Caleb Gowan – \$2,500

Addison Cox – \$864

Hadley Alexander – \$784

Oliver Britten – \$500

Thank you from the bottom of our hearts.

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

Totals for year ending 12/31/13

WINTER, SPRING, SUMMER OR FALL, THE STURGIS FAMILY STAYS VERY BUSY!

DAY OF HOPE AND NATALIE'S WISH CELEBRATION

We recently returned from the CRF Day of Hope Family Conference. First, we must thank Nancy, Jeff and Natalie Stack for helping cystinosis families get to the conference and for making everything so easy and enjoyable once we were there. The conference changes the lives of everyone who attends it. It is a magical coming-together and uniting as a family. When the conference is over, each family returns to their homes – and each of them, in their own unique way, has gained the renewed encouragement, motivation and inspiration they need to help them in their daily lives until we find the cure.

One special highlight of the event: Erin Little made a wonderful keepsake for Nancy and presented it to her as the conference ended. Using fabric scraps sent to her by families from around the country, the quilt was a replica of the Day of Conference tree logo. It was a beautiful, truly one-of-a-kind gift for a remarkable, one-of-a-kind person who enriches all of our lives.

I'm thankful to everyone who helps in the fight against cystinosis. There is no more wonderful time to experience all the goodwill and generosity our community receives than at the annual Natalie's Wish Celebration. The energy and love were palpable at this year's event.

Thank you to all of the attendees and donors who have joined us on our journey.

Henry enjoying himself at the Day of Hope Family Conference at the Balboa Bay Resort in Newport Beach, California

(Photo next page, lower right) Henry with his teacher, Mrs. Dash, and two classmates at his ski event

SCHOOL NEWS

Henry is doing well right now. He loves playing *Star Wars*, and his favorite foods haven't changed much. For dinner, he still loves steak, corn with lots of butter and salt, and ice cream. It's usually eggs with Frank's Red Hot Sauce for breakfast, and lunch includes a turkey sandwich with mayo on a hoagie roll, and a chocolate chip cookie, especially if Grandma Sturgis happened to bake them.

Henry is finishing first grade with his teacher, Mrs. Dash – who he loves. Mrs. Dash donated proceeds from her annual Halloween party to 24 Hours for Hank. We can't thank her enough for all she has done for Henry this year!

We are also grateful for the other teachers and therapists who work with Henry. They have all played a role in Henry's success this year and they should all celebrate his achievements.

Farmin Stidwell, Henry's school, also deserves a "shout-out." Students selected 24 Hours for Hank as their charity for "Cents for Cystinosis" – a fundraiser put on by the Jogging Club. The school also had a four-person team in our ski event. Henry's class chose the name: "Jedi Snow-Storm" – yes, we are into *Star Wars* right now. I was honored by how many teachers volunteered at the event.

Recently, the physical therapist was testing another classmate's eyes, so she decided that she would also test Henry. Sure enough, he showed signs of "convergence deficiency," which could be hindering his reading ability, as well as causing his headaches.

Henry has a mild form of convergence deficiency. Therapy is available but it is extremely expensive and it's two hours from our home. We've opted for a home therapy: Computerized Vergence Exercises. We haven't started yet, but plan to do so soon. If anyone has questions, I'd be happy to share information. (tricia.sturgis@gmail.com)

Henry uses removable orthotics in his shoes to keep him balanced and stable as he moves. We're working with Mary Fischer, a professional who runs ADAPT (Abilities are Determined by Attitude Perseverance and Teamwork) in Coeur d' Alene, Idaho. It is a fitting acronym for children with cystinosis.

SUMMER VACATION

Our summer plans involve outdoor adventures, boating around the lake, attending camp, gardening and traveling to the Oregon Coast.

**In the chaos of everyday life,
remember to enjoy the little things.
Tricia Sturgis**



*Photo courtesy of Bob Hodson Photography
At the Natalie's Wish Celebration in April,
Henry and his parents, Tricia and Brian Sturgis,
presented a check to CRF for \$120,000.*

MEDICAL NEWS

When school is out in June, we will be heading to Henry's annual appointment at Seattle Children's Hospital. He will see four specialists and a metabolic dietician. We always learn a great deal when we go, and make great strides in Henry's overall care. This year, we hope to explore the goal of swallowing small pills, which could lead to the bigger goal of removing Henry's G-tube.

Henry continues to use Procsybi® and the Cystaran™ eye drops. His blood/chemistry levels have been great.

In March, I attended CRF's Fourth International Research Symposium. Sixty renowned scientists and doctors from around the world shared their latest research successes, giving me and the other parental attendees great hope for the future. I am genuinely impressed by the researchers' efforts and appreciate everything they do for our children.



Cowboy Up



for a Cure

More than 110 skiers and snowboarders ages 5 to 73 from Sandpoint, Idaho and the surrounding region – and as far away as Arkansas, Iowa, California, Oregon, Colorado and Washington, D.C. – participated in the *24 Hours of Schweitzer* ski event on March 21 and 22. With a theme *Cowboy Up for a Cure*, the round-the-clock relay was a huge success.

Bolstered by sunny skies, good weather and the generosity of the Sandpoint community, participants raised more than \$100,000 at one of the largest cystinosis events in the country. That brings the 24 Hours for Hank Foundation's fundraising total to over \$725,000 in six years.

The event concluded with a lively auction, dinner and awards party attended by 230 people. Nearly 150 businesses and individuals contributed to the auction dinner and awards party, including more than 125 live and silent auction items and prizes.

"We are amazed by the skill, endurance and spirit of participants, young and old – and their unwavering dedication to our cause. We'd like to thank everyone who helped create another amazing event – participants and their families, donors, volunteers, sponsors, supporters, area businesses, local media, Schweitzer Mountain Resort and its employees, and our family and friends," said Brian Sturgis, Hank's dad. "Hank and others around the world are already benefiting from new treatments developed with money raised on Schweitzer Mountain."

Finally, we want to thank our 24 Hour Board members and the community of Sandpoint, as well as our amazing title sponsors: Schweitzer Mountain Resort and Subaru of North America. We wouldn't be where we are today, without you!

All money raised through 24 Hours for Hank is sent to the Cystinosis Research Foundation. The foundation's costs are 100 percent underwritten, so every dollar donated goes directly to CRF-funded scientists at leading universities and medical centers in the United States and around the world.



**Special thanks
to our fantastic
Title Sponsors**





THE MOST **IMPORTANT** SURVEY YOU MAY EVER COMPLETE

We are constantly asked to complete surveys, so you may find yourself asking why you should participate in the yearly CCIR survey.

The bottom line is that CCIR works for the cystinosis community. The medical history you provide chronicles the progression of the disease, and it helps get the message across that much still needs to be done to improve quality of life.

There is no cystinosis database of this scale in the world and surely none that makes the anonymous information freely available to those with the potential to change the course of the disease forever.



The CCIR is available in English, French, Spanish, Dutch and Portuguese. Register at

www.cystinosisregistry.org

HOW CAN A SURVEY CHANGE ANYTHING?

Chances are that on more than one occasion you have had to explain what cystinosis is. You may even have had to explain it to a physician who has never encountered a case before. The truth is that your first-hand experience with the disease makes you a cystinosis expert.

When you share your experience, we have a better chance to:

- Improve diagnosis of this rare disease
- Improve access to established treatments
- Help people understand that cystinosis patients have unmet needs
- Gain support for further research and drug development



DON'T WE ALREADY KNOW A LOT ABOUT CYSTINOSIS?

Believe it or not, despite the advances made in cystinosis in the last 50 years, there is still much we do not know about it. This is due in part to how difficult and expensive it is for researchers to study and track rare disease populations. Frequently, we do not know the actual occurrence of the disease. CCIR is the best system currently available to identify and track cystinosis cases worldwide. However, CCIR's success relies on having both patients and their caregivers on board, so we need your help to model participation and spread the word to the community.

WHY DO WE NEED ANOTHER CYSTINOSIS SURVEY?

CCIR was created primarily to circumvent stalls in the drug-development process that are often attributed to lack of understanding of the natural history of rare diseases. And since cystinosis is a global disease, a large-scale effort is needed to capture the full scope of the disease's impact. But cystinosis surveys have been done before, so perhaps you are wondering why yet another survey is necessary. The fact is that not all surveys are the same.

Surveys can vary in the following ways:

- Surveys have different goals and are used for different purposes.
- Survey information is recorded in different formats and in multiple places.
- Access to data is often limited and not openly shared with the general research community.

CCIR allows anyone with a legitimate research interest in cystinosis to access a central, secure database, and take away whatever anonymous information they need to advance cystinosis projects.

WHAT WILL YOU GET OUT OF PARTICIPATING?

An opportunity to advance therapies for cystinosis by simply spending 30 minutes a year completing and updating the survey in the privacy of your own home.

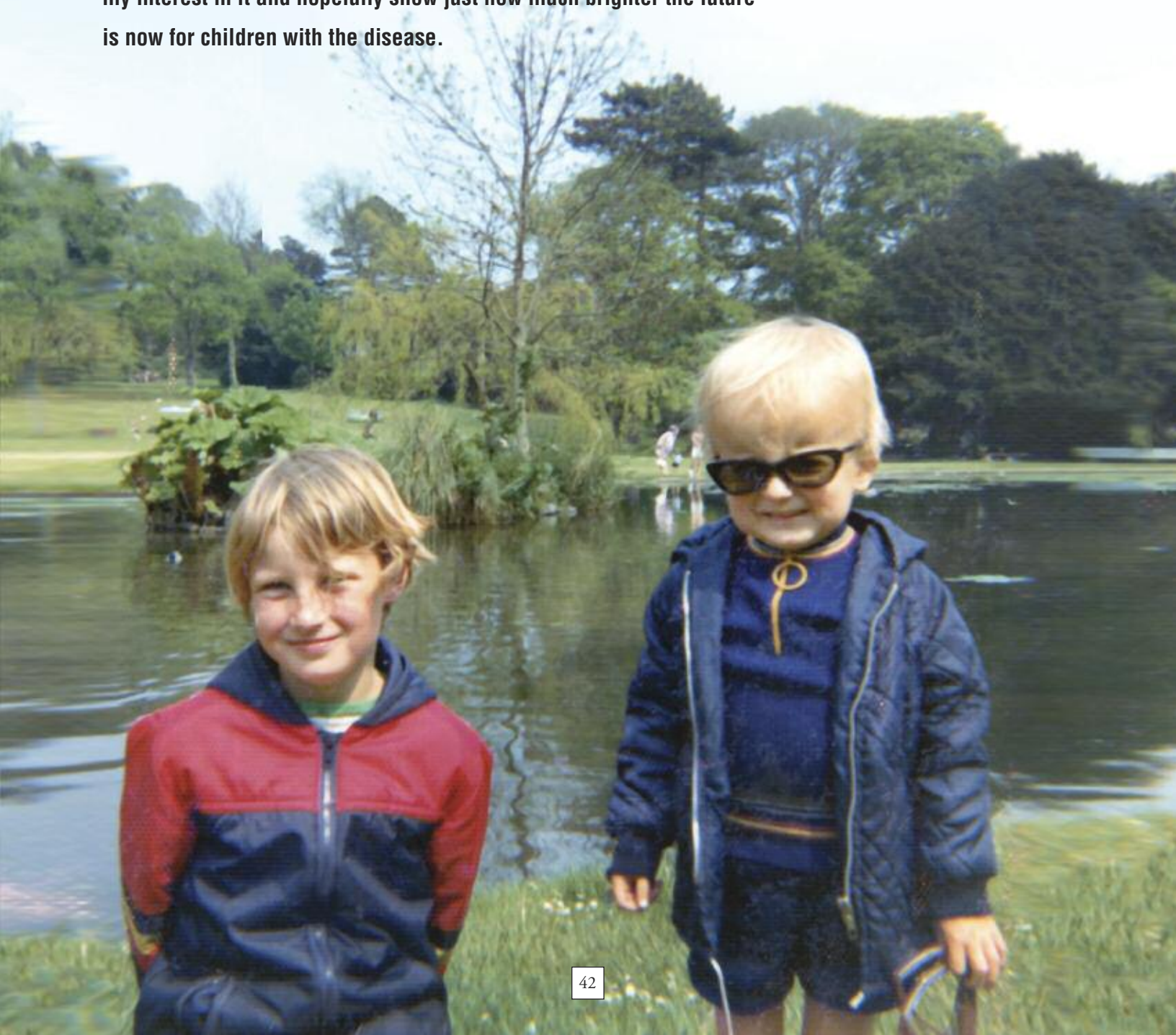
- Direct access to registry data so you can see for yourself what cystinosis is shaping up to look like across the globe.
- Access to medical experts who will answer your questions about the disease. (See the *Ask an Expert* feature at www.cystinosisregistry.org.)
- Notifications of upcoming clinical trials and important cystinosis news updates.
- Pride in knowing that cystinosis is one of fewer than 20 percent of rare diseases that has a dedicated registry.
- A sense of belonging to a CCIR community that is 440 people strong and has representation from 39 different countries.

A rare look at living with cystinosis 45 years ago

In Loving Memory of **DAVID JARVIS BANHAM**

*By Julia Wells, David's mom,
Stirling, Ontario, Canada*

My name is Julia Wells and I am the mother of a child who was born with cystinosis. While I have only been aware of the Cystinosis Research Foundation (CRF) for a matter of months now, my story will illustrate my interest in it and hopefully show just how much brighter the future is now for children with the disease.



My son David Jarvis, was born in 1969, a nine-pound healthy baby and a beloved brother to his three-year-old sister, Rachel. For the first couple of months he was an easy to feed, contented baby who slept well, but around three months old that changed. He started to throw up, becoming a fussy baby who did not gain weight.

Over the next few months blood and urine tests were done by the family doctor, diabetes was considered as a possibility, but then discounted. Then we were referred to a pediatrician. At some point, the term Fanconi syndrome was talked about and medication was prescribed, but since most of the time he either couldn't get it down, or threw it back up, it didn't really help. This continued for several months with no real diagnosis for us as parents to hold onto, and the daily worry of how to feed him and get his medication into him was very stressful.

After about a year of regular unsatisfactory visits to the pediatrician, David's condition was not getting any better, and my frustration level was very high with the worry of him not eating and constantly vomiting. During one visit, I asked how was he ever going to improve if he couldn't keep any of the medication down. I was bluntly told that his illness was terminal and his life span would be short. I walked away from that appointment horrified at the news that my worst fears had been realized. Looking back I find it hard to believe that no assistance of any kind was offered to help his father and me deal with this devastating news.

We found it hard to get information from our pediatrician about what to expect. For example, it was only after we noticed his eyes

all gummed up after a sunny day that we were told about the eye problems and to keep him out of the sun.

Also, it wasn't until we saw another pediatrician, who was on duty during one of our visits, that we were told it was a genetic disease and that there was a one in four chance that any child we had could have the disease. I was only 23 at the time and this was key information because we had planned to have more children. That news changed everything.

but was very aware that they were not going to prolong his life. Our focus was to make sure that he had as normal a life as possible, because although frail physically, he was a bright, fun-loving little boy.

At about two years of age he started to eat small amounts and vomited less, but he never gained any more weight, had bent legs from the rickets, and wore dark glasses whenever outside to try and protect his eyes.



Soon afterwards we moved to another part of England and were put in touch with a different pediatrician who referred us to a kidney specialist in one of the teaching hospitals in London. During the next couple of years they did everything they could to help make David's life as good as possible.

After all this time I honestly can't remember the names of the 11 drugs and supplements he was on,

Although small for his age, he loved to play outside with Rachel and his friends, and when his limited energy gave out, he sat on his little tricycle which made it possible to spend more time out there with them. At the age of five he started school and was very proud of his school uniform, particularly the school cap. While the principal had at first been reluctant to enroll him, because he was concerned about



his safety, the other children were amazingly kind to him. They made sure that he was safe outside at recess and would kick his ball back to him so he didn't have to run around too much. He also had Rachel there too, and she was very good at looking after him.

Our beloved David died at the age of five years and three months in August 1974. From the time he was born, until very recently, I had never known anyone else who had a child with cystinosis.

FROM THE TIME DAVID WAS BORN IN 1969, UNTIL VERY RECENTLY, I HAD NEVER KNOWN ANYONE WHO HAD A CHILD WITH CYSTINOSIS.

All this changed in 2009 when I saw an article in our local paper that caught my attention. The article was about a little girl named Gabbie who had cystinosis, and whose grandparents lived locally. I was very interested to read the article, and about the advances that had been made, but I didn't see any reason to get in touch because I didn't feel I had anything helpful to share with them.

Then a couple of months ago, there was an ad in the local paper for a fundraising dance in aid of cystinosis, which was to be held in a neighboring town. This time I felt I had to get in touch to find out more and discovered that it was the best friend of Gabbie's mum, who was organizing the event. Now, I felt there was something useful I could do.

I sent an e-mail to everyone I knew who might support the dance, either by attending or sending a donation. My friends rallied round and those who couldn't attend the dance sent in donations. About a dozen were able to join my husband, Rachel and I at the event, where, we were fortunate to meet Jody and her husband, Trevor.

Jody gave me a copy of *Cystinosis Magazine* and I was fascinated to read about of all the amazing research being done. The stories of the different families often brought me to tears as I could appreciate and empathize with so many of the issues they are dealing with.

Where I had felt completely alone and isolated throughout David's short life, and indeed ever since, I found that families now have a network of people through CRF.

After the fundraising experience and reading the magazine, I feel a connection to a large group of people in Canada and the United States. With the magazine, I can keep in touch with

what is happening both on the research front and with the families. At some point I'm sure I will be able to help in another fundraiser.

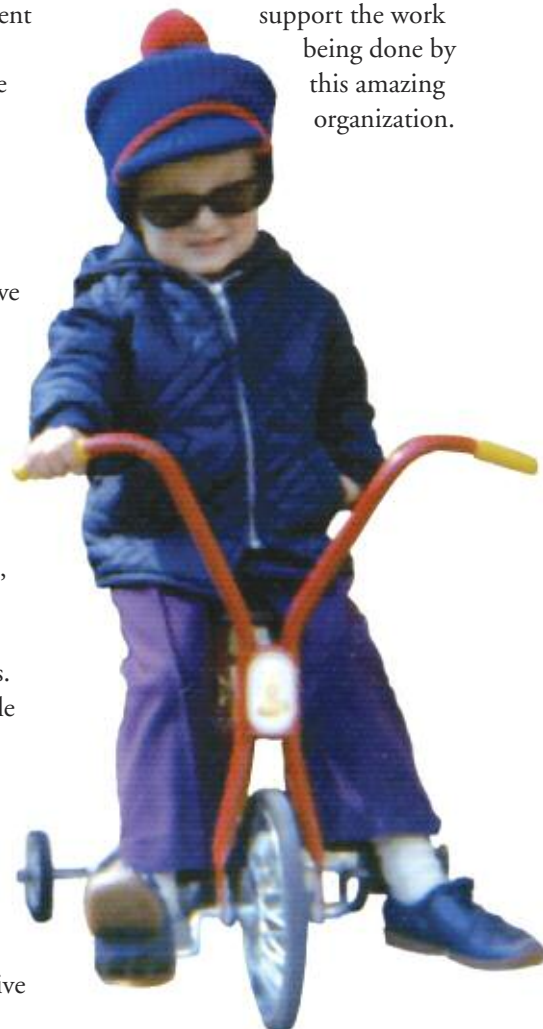
Things have certainly changed since my beloved little David was alive. The advances in drug therapies, the ground-breaking research, the sense of commitment and community spirit within CRF, and the extensive fundraising, all give

great hope that it won't be long before a diagnosis of cystinosis is no longer the heartbreaking news that it was when David was born.

As I listen to Jody talk about all the research being done, her ability to talk directly to the researchers, and having the support of everyone through the CRF and its wonderful magazine, I realize how different it is now. There is so much community spirit within CRF, so much better quality-of-life for the children now and so much more hope that a cure will be found.

I am so glad that I saw the ad for the dance and got to meet Jody. Through her I now have a connection to CRF. I feel as if Rachel and I have become honorary members of a club we never knew existed.

I look forward to finding more opportunities to help support the work being done by this amazing organization.



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Whitney Glaize with her fiancé, Jason McElroy

Whitney's Perspective: Her Kidney Transplant

Perhaps one reason my transplant was so difficult was that my body had become used to cystinosis and I functioned very well with it. I was 33 when I was transplanted so my body had many years to become accustomed to life with cystinosis.

Some of the most severe post-transplant complications, are the BK virus and antibodies. The BK virus showed up about two years after my transplant. This virus is virtually harmless in individuals with normal immune systems and is so common that doctors don't test for it pre-transplant (it is estimated that about 80 percent of the population has the virus). In individuals with kidney transplants the virus can cause kidney damage and ultimately cause the transplanted kidney to shut down.

Treating BK is not an exact science. What works for one patient does not necessarily work for another. It is imperative to get the virus under control as quickly as possible; otherwise the transplanted kidney is at risk. We have had quite a bit of success in reducing my anti-rejection medications and dosage.

My nephrologist still can't believe how little anti-rejection medication I need. For a transplant patient, I am on a very low dose of anti-rejection medication and I am pushing for even further reductions. I've experienced many negative side effects from the medications that suppress the immune system.

If I had it to do over again I would find a transplant program that does not use medication but the donor's immune system producing stem cells to protect my body from rejecting my mother's kidney. I hate the anti-rejection medications I take. I went through several combinations before finding the cocktail that my body would tolerate.

My body is beginning to develop antibodies against my mom's kidney, which means my immune system has detected somebody else's organ inside of me and is shutting down the "new" kidney. The typical treatment is more anti-rejection medication. However, in someone with BK, more anti-rejection medication means risking multiplying the virus and the possibility of damaging the transplanted kidney. So the question has been: do I lower my medication to treat BK or increase it to treat the antibodies? Lowering the medication worked best for me but finding the right balance has been stressful. I think my poor

I am officially 2 years and 5 months post-transplant. And I think things have finally settled down and I can breathe a sigh of relief. I didn't expect so many ups and downs after my kidney transplant on October 26, 2011.

The doctors painted a slightly unrealistic picture of what life would be like after I received my mom's kidney. I thought once I recovered from the actual surgery I would be good to go. The medical team failed to tell me that there are many complications with having a kidney transplant. Unfortunately, I've had to deal with a lot, if not most, of them!

*By Whitney Glaize
Orlando, Florida*

nephrologist wants to retire after dealing with me. He always says, “It’s always an adventure with you Whitney,” which I take as, “You are one of my most challenging patients Whitney.”

If I could trade places with someone who has not had a kidney transplant, I would do so. I thought my life would be easier than before the transplant, but I was wrong. Life is actually more difficult. I take more medicine now and I take all the same electrolytes, minus one, phosphorus – the only medicine the transplant eliminated from my regiment. I still drink more than 100 ounces of water each day because Fanconi syndrome is still alive in my native kidneys, although we keep hoping they will shut down one day.

HAVING A TRANSPLANTED KIDNEY IS NOT EASY. MY “NORMAL” HAS JUST CHANGED, MUCH LIKE A RECENTLY DIAGNOSED FAMILY’S CHANGES WHEN THEY GET THEIR CHILD’S DIAGNOSIS.

And, I get my blood drawn more often now than pre-transplant. Before receiving a new kidney we knew my kidneys were going to fail, we just didn’t know when (actually my pediatric nephrologist, who I credit with saving my life, predicted he would be retired by the time I needed a new kidney and he was right). Now rather than expecting my kidney to fail, we are continually monitoring to make sure that it doesn’t.

I feel good though – better than I ever have!

Having a transplanted kidney is not easy. My “normal” has just changed, much like a recently diagnosed family’s changes when they get their child’s diagnosis. I do an amazing job of disguising the symptoms, but I still have cystinosis. I still take Cystagon®, use the eye drops, and I work hard to avoid the complications cystinosis causes. And I believe I am succeeding.

I work full-time as a special needs preschool teacher, and I run a speech therapy business on the side. I work out and weight train weekly. I keep a busy social calendar. Perhaps my nephrologist of the last 10 years summed it up best when hearing news of my recent engagement, “Maybe Jason can slow you down.” Not a chance Dr. Warren, not a chance.



In Memory

Nathaniel Wagler, dear friend and longtime member of the cystinosis community, passed away on May 4, 2014 at Listowel Memorial Hospital in Ontario, Canada.

Nathaniel was the first of five children born to Elroy and Diane Wagler on July 22, 1980 in Listowel, Ontario. He was a strong healthy baby until he started to have unexplainable health issues at four months. In 1981, after many months of being sick without medical explanation, Nathaniel was diagnosed with cystinosis, just months before his brother Timothy was born on August 5. In September 1982, Timothy was diagnosed with cystinosis.

Elroy and Diane Wagler have three healthy daughters, Anita, Lynette and Loretta, who are in their twenties and thirties.

Nathaniel had a big heart and was loved by everyone who knew him. He worked hard in the family business until he grew too weak to do so, but as he lost strength he continued to train his dogs and horse. Throughout his 33 years he lived life fully and he never gave up hope.

Cystinosis Magazine published an article about the Wagler family in its spring 2012 issue. That article is available at <http://www.cystinosisresearch.org/events-news/newsletters/>

In lieu of flowers, the family requests that donations may be made to the Cystinosis Research Foundation or the Masters International Ministries.



Photo courtesy of Lars Wanberg

'Now He's Really Like My Brother'

By Dennis Arp

An extraordinary donation not only gives life, but it also intensifies the connection between two cystinosis families.

On the surface, it was just a phone call from a friend. But at its heart, it was “the most beautiful, amazing thing ever.”

It was October 2013, and Jessica DeDio was at UCLA Medical Center with her older son, Bailey, who has cystinosis. They were waiting for Bailey’s profile to go live on the cadaver kidney donation website as Jessica’s friend Tanya Chilcott updated the Bailey Believes – Donate Life page on Facebook. The youngster’s kidneys were failing, and social media was a way to alert friends about Bailey’s urgent need for a transplant and to share his blood type.

Within minutes, Jessica got a call from Mary Jordan, whose son also has cystinosis.

“Jessica, I have O positive blood,” Mary said excitedly. “I can be Bailey’s donor.”

Just like that – no qualms, no hesitation.

Who knew that a family friendship born of a shared health crisis could suddenly vault to angelic heights?

Jessica and Mary had met several years before at Stanford, where their sons were both participants in a study of the delayed-release version of cysteamine, the drug commonly used to treat cystinosis. Bailey and Joey hit it off immediately. “They’re almost like twins,” Jessica said. “Same red hair, same interests.”

The two families also bonded, and they started coordinating their trips to Palo Alto for the boys’ treatment. Over the years, the two moms supported each other through the many ups and downs of cystinosis treatment.

The disease attacks all organs of the body, especially the kidneys, but it was still a blow to everyone when

Joey started suffering renal failure a few years ago. Mary went through all the testing and was deemed a healthy candidate to donate a kidney to her son, but as it turned out the blood markers were better with a cadaver organ, so that’s the kidney he got.

Jessica was there for Mary as Joey went through a year and a half of organ rejection, and then after the youngster turned a corner, Bailey’s need for a transplant suddenly became acute. Jessica had long planned to donate to her son and so was heart-broken when she learned that she wasn’t a match.

“I love Bailey, Jessica, Jay and little Ryder,” Mary said of the entire DeDio family. “When I learned that no one was matching, and that Bailey was going on dialysis, there was no question that I would donate if I could. I went home and told my husband and others, and they said to go for it.”

More testing at UCLA confirmed Mary as a qualified donor, and on Jan. 14 surgeons removed one of her kidneys and transplanted it into Bailey.

After the surgery, Jessica and her family went to see Bailey in the OR recovery area, and she asked the nurse if she could also see Mary. A groggy but unmistakable voice came from two gurneys down.

“Mary was smiling,” Jessica recalled. “It was a smile that only a true angel would have. Her first words were ‘How’s Bailey?’ That’s so like Mary – never about her, always about Bailey.”

Mary deflects any notion of personal heroism.

“Bailey and Joey are the real heroes of this story. Now they’re both doing terrific, and that’s what counts. It feels good to know I had something to do with that.

“I’m a strong practicing Catholic, and it makes my faith stronger that this was able to happen. It makes me feel better inside my heart, and it makes my family feel better – that it’s

not just talking about helping other people, it's doing it."

The focus stays right where it belongs, she added. "Bailey is alive. Joey keeps his friend."

And what friends they are, these two high school students – Bailey in Fullerton, in Southern California, and Joey in Rocklin, in the state's northern Gold Country.

"When they're together, they're inseparable," Mary said. "They're on the computer together, they play together. It's like they have the same personality."

And now, the links between the two families are even stronger.

"I do feel like I have a special bond with Bailey, because he has a part of me in him," Mary said. "One time Joe said, 'Now he's really like my brother.'"

For Jessica and Mary, the connection crosses all boundaries.

When they speak, it's in a shorthand and with a depth of shared experience that acknowledges the 85 meds a day, the hourly eye drops – the needles and the nausea, the pain and the fortitude.

"I have lots of friends, and she has lots of friends," Mary said. "My friends kind of get what cystinosis is. I know that Jessica really gets it."

Mary hopes that others in the cystinosis community and beyond will take the time to get tested and consider getting on the donor registry. "You only need one kidney," she said. "I don't even notice the difference."

Jessica sure notices the difference in Bailey. There were some tough times right after the surgery, as he battled a virus and adjusted to new

meds. But now "he's so much better than before," Jessica said.

When Bailey went on dialysis, she worried that he might never again experience some of the best parts of his life – like riding his beloved dirt bike. Bailey got a motorized bicycle

for Christmas, as kind of a transition vehicle, "and to see him pedal that bike up and down the street, when before he had days when he didn't want to get out of

bed, well, it's just wonderful to see," Jessica said.

Now Bailey can look forward to Nov. 8, when he plans to participate in the third annual Bailey Believes Ride for a Cure. The fun-filled day of dirt-bike riding is followed by a night of food, raffles and an auction that raises thousands of dollars for the Cystinosis Research Foundation. **Visit www.cystinosisresearch.org/donate-for-bailey.**

"We want to be part of finding a cure," Jessica said. She also wants her friend Mary Jordan to know just how much she means to her.

"It's really hard for me to find the perfect words to express my gratitude," Jessica said. "And it's not just Mary. Her whole family (husband Richard, daughter Caitlin Marie, sons Joseph, Patrick and Connor) had to do without her for a while.

"Mary is the most selfless, loving, wonderful person, and her donation is the most beautiful, amazing thing ever. I feel so blessed to know her."

Jessica calls the whole dialysis-to-transplant experience traumatic "and yet also so full of compassion and love. It's so hard for me to find the right words."

So she turns to a phrase that Bailey would appreciate.

"It's been quite a ride," she says.

BAILEY AND JOEY ARE THE REAL HEROES OF THIS STORY. NOW THEY'RE BOTH DOING TERRIFIC, AND THAT'S WHAT COUNTS.

Bailey with his transplant donor Mary Jordan – and Joey's mom.



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



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Cystinosis Research Foundation received a generous \$20,000 donation from the Share Our Wine Foundation's annual wine auction attended by wine enthusiasts from around Orange County.

The event, sponsored by South Coast Plaza, benefitted children's charities in Orange County. CRF would like to thank event supporters, the Share Our Wine Foundation board and its president Danielle Staffieri, for their generosity.



Trick or Treat ... for A Cure

The Third Annual Lots of Love for Landon Halloween Party, hosted by Lauren and Jimmy Hartz in honor of their son, Landon, was held on Saturday, November 2, 2013 and drew more than 300 partygoers – its largest crowd ever.

The evening's festivities included a Chinese auction, door prizes, dinner, costume contests, plus a live DJ who entertained the large crowd. **The event raised an incredible \$10,000 for cystinosis research!**



These Regulars are Anything But

Nicole and Brad Manz, one-year-old Keegan Manz's mom and dad, were approached by a local group known as *The Regulars* who fundraise in their community of Cary, North Carolina. *The Regulars* are comprised of local bands who perform to help people in need in their community.

After meeting with Nicole and Brad, *The Regulars* told them that the money raised could be used for whatever the Manz family wanted – a vacation or to pay bills. The Manzs' priority is to support research for better treatments and a cure for cystinosis so **they presented the \$4,500 donation to Cystinosis Research Foundation in Keegan's honor.**

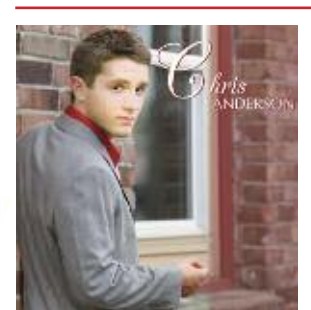
Thank you to *The Regulars* and the Manz family for joining CRF's quest for a cure!



HIGH SCHOOL STUDENTS HOOP IT UP FOR A CURE

The Clarkston High School Bantams of Washington and The Lewiston High School Bengals are fierce crosstown rivals, but once a year they join forces to support local charities through their spirited Golden Throne Week activities. In January, teams from both schools played like champions in the 20th Annual Golden Throne basketball games to gain control of the coveted Golden Throne trophy.

But the real winner was 10-year-old Tina Flerchinger from Clarkston. Her foundation, Tina's Hope for a Cure, was one of three lucky charities to benefit from this year's festivities. **Tina's Hope for a Cure received a check for \$3,561 from the high school students, who played their hearts out to raise money for cystinosis research.**



The second annual benefit concert, *Music for Mary*, was held on Saturday, March 22 in honor of five-year-old Mary Head from Yelm, Washington. Talented singer and local favorite, Chris Anderson performed after guests enjoyed an authentic Italian dinner prepared by Viafores Italian Delicatessen. Guests then had a chance to participate in exciting silent and live auctions.

The event, which took place at the Tacoma Sportsmen's Club, raised more than \$17,890 for cystinosis research. Thank you to Melissa and J.R. Head, their family and friends who work throughout the year to help find a cure for cystinosis.



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BAILEY FEST 2014

Saturday, August 9, 2014
Bailey Fest 2014 – Bailey DeDio
 SeaHawk Cocktail Lounge
 Lakewood, CA
Information: Tanya Chilcott
 (562) 858-6796

Friday, October 3, 2014
A Night of Hopes & Wishes
 Jake Krabe, Medina, Ohio
Information and tickets:
www.cystinosisresearch.org/donate-for-jake



Saturday, September 13, 2014

Third Annual Fore Fathers Memorial Golf Tournament
 Andrew Cunningham



Boulder Creek Golf Course, Landon, Alberta, Canada
Information: Neil McCullagh (403) 835-7593
 or neilmccullagh@gmail.com



Saturday, November 8, 2014
Ride for a Cure – Bailey DeDio
 Barstow/Stoddard Valley, CA
Information or tickets:
www.cystinosisresearch.org/donate-for-Bailey



Saturday, September 20, 2014
Swing, Shoot and Liv Golf Classic
 Olivia Little – Saugeen Golf Club, Port Elgin, Ontario, Canada
Information: Erin Little
 (519) 832-5188

Coming March 2015
Seventh Annual 24 Hours of Schweitzer
 Henry Sturgis
 Schweitzer Mountain Resort
 Sandpoint, Idaho



Monday, October 27, 2014
 Seventh Annual Natalie's Wish
 Fore a Cure Golf Tournament
 Santa Ana Country Club
Information: Zoe Solsby
zsolsby@cystinosisresearch.org

April 16 – April 18, 2015

CRF Day of Hope
 Family Conference
 Newport Beach, CA

Information: Nancy Stack
nstack@cystinosisresearch.org



The Year's Biggest Celebration
The 2015 Natalie's Wish Gala • Saturday, April 18
 Newport Beach, California • **Information: (949) 223-7610**



COLLABORATION, ADVANCES HIGHLIGHT FOURTH CRF INTERNATIONAL CYSTINOSIS RESEARCH SYMPOSIUM

By Dennis Arp

After two incredibly full days, rich with recent findings, keynote addresses, panel discussions, poster sessions and new opportunities for collaboration, it was Dr. Julie Ingelfinger's task to try to sum up the Fourth International Research Symposium held March 6–7 in Irvine, California.



Julie Ingelfinger, MD



Ana Maria Cuervo, MD, PhD

Starting with some historical perspective, Dr. Ingelfinger, a renowned pediatric nephrologist and early clinician in the treatment of cystinosis, observed that “one can see a lot of bad things happen when cystinosis goes untreated. But now people are living longer, and a lot of what was presented here provides hope for the future.”

Hosted by the Cystinosis Research Foundation, the symposium brought together more than 50 researchers and clinicians from all over the globe – each with a special expertise in some aspect of the care and treatment of cystinosis.

The symposium is held every two years at the Arnold and Mabel Beckman Center of the National Academies of Science and Engineering, and each event has helped cultivate an atmosphere of sharing and synergy that speaks to the collegiality within the cystinosis research community. The most recent meeting was no exception.

The symposium was kicked off by a keynote address by Dr. Ana Maria Cuervo, who is on faculty at the Albert Einstein College of Medicine in New York City. Dr. Cuervo is a leader in the study of autophagy, a field that may provide building blocks that can fuel continuous cell survival.

“Dr. Cuervo’s presentation and interactions offline during the meeting are likely to lead to important collaborations, and I think that will be very worthwhile to watch,” Dr. Ingelfinger noted.

More than 15 other talks and formal presentations followed Dr. Cuervo’s keynote, providing insights to the wide array of important investigations, all made possible by funding from the Cystinosis Research Foundation.

“So what does all of this mean?” Dr. Ingelfinger asked during her summary talk on the final day. “I think it means that as there is better understanding of the different cystinosis partners and pathways, there will be coordinated therapy and new ways to use multiple drugs and ultimately perhaps a cure.

“I think the collaborations fostered here are invaluable. And as someone who has a lot of experience with cystinosis, I find it very exciting that there are so many young investigators with us today.”

In wrapping up the conference, Dr. Ingelfinger pointed to the \$25.8 million raised by the CRF and said, “It has changed the treatment of this disease and will change it more in the future.”

Looking into that future, she envisions not just a cure for cystinosis but advances in basic science that transcend the condition, as well as additional collaboration among investigators, patients, caregivers, clinicians and scientists.

In other words, she concluded, “a true opportunity for translational medicine.”

CRF’s research funding has provided “a true opportunity for translational medicine.”





A patient support program for PROCYSBI® (cysteamine bitartrate)
delayed-release capsules

COMMONLY ASKED QUESTIONS ABOUT PROCYSBI®

Q: What can I expect when I start PROCYSBI?

A: As with any new medicine, it may take time for your body to adjust to PROCYSBI. It is important to work with your doctor to find the best way to take PROCYSBI that works for you. For example, your doctor may start you on a lower dose and adjust it over time to a level that suits you best.

Q: Can I take PROCYSBI with water?

A: Yes, you can take PROCYSBI with water when swallowing your capsules whole.

Q: Is it ok to take PROCYSBI with food and beverages?

A: It is ok to take PROCYSBI with or without food and beverages. However, it is very important to take PROCYSBI with a similar type and amount of food every time, and to check in with your doctor as you are adapting to your new medicine.

Q: How should I take PROCYSBI if I'm using a Feeding Tube?

A: PROCYSBI can be administered through a G-tube (or feeding tube). Please contact a RaptorCares nurse before starting PROCYSBI with a feeding tube or ask your doctor about a video showing G-tube administration.

Q: What if I or my loved one have side effects?

A: With any side effect, you should notify your doctor right away. Your doctor may lower your dose and then gradually increase it to achieve your target dose. Remember not to stop taking PROCYSBI or any medication without talking to your doctor first.

Q: What financial support programs are available to help me?

A: Co-pay assistance, secondary insurance, travel, and white blood cell cystine testing support are available to eligible patients. The first step is always to contact RaptorCares at 855.888.4004.

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Please check the Patient Package Insert that comes with your PROCYSBI regarding these topics as well as other helpful information.

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IMPORTANT SAFETY INFORMATION

What is PROCYSBI (Pro-CIS-bee)?

PROCYSBI is a prescription medicine used to manage a medical condition called nephropathic cystinosis in adults and children 6 years of age and older.

It is not known if PROCYSBI is safe and effective in children under 6 years of age.

Who should not take PROCYSBI?

Do not take PROCYSBI if you are allergic to penicillamine.

What should I tell my doctor before taking PROCYSBI?

Before you take PROCYSBI, tell your doctor if you:

- have a skin rash or bone problems
- have a history of seizures, exhaustion, sleepiness, depression, or other nervous system problems
- have or have had stomach or bowel (intestinal) problems including ulcers or bleeding
- have liver or blood problems
- are pregnant or plan to become pregnant. It is not known if PROCYSBI will harm your unborn baby. Tell your doctor right away if you think that you are pregnant. Talk with your doctor about the benefits and risks of taking PROCYSBI during pregnancy.
- are breastfeeding or plan to breastfeed. You should not breastfeed during treatment with PROCYSBI. Talk with your doctor about the best way to feed your baby if you take PROCYSBI.

Tell your doctor about all medicines that you take, including prescription and over-the-counter medicines, vitamins, and herbal supplements.

Know the medicines you take. Keep a list of them to show your doctor and pharmacist when you get a new medicine.

What should I avoid while taking PROCYSBI?

Do not drive or operate heavy machinery until you know how PROCYSBI affects you. PROCYSBI can make you sleepy or less alert than normal.

What are the possible side-effects of PROCYSBI?

PROCYSBI can cause serious side-effects, including:

- **Skin, bone, and joint problems.** People treated with high doses of cysteamine bitartrate may develop abnormal changes of their skin and bones. These changes may include stretch marks, bone injuries (such as fractures), bone deformities, and joint problems. Check your skin while taking PROCYSBI. Tell your doctor if you notice any skin changes. Your doctor will check you for these problems.

- **Skin rash.** Skin rash is common with cysteamine bitartrate and may sometimes be severe. **Tell your doctor right away if you get a skin rash.** Your dose of PROCYSBI may need to be decreased until the rash goes away. If the rash is severe, your doctor may tell you to stop taking PROCYSBI.
- **Central nervous system symptoms.** Some people who take other medicines that contain cysteamine bitartrate develop seizures, depression, and become very sleepy. The medicine may affect how your brain is working (encephalopathy). Tell your doctor right away if you develop any of these symptoms.
- **Stomach and bowel (intestinal) problems.** Some people who take other medicines that contain cysteamine bitartrate develop ulcers and bleeding in their stomach or bowel.
- **Tell your doctor right away** if you get stomach-area pain, nausea, vomiting, loss of appetite, or vomit blood.
- **Low white blood cell count** and certain abnormal liver function blood tests. Your doctor should check you for these problems.
- **Benign intracranial hypertension** (pseudotumor cerebri) has happened in some people who take immediate-release cysteamine bitartrate. This is a condition where there is high pressure in the fluid around the brain. Your doctor should do eye examinations to find and treat this problem early.
- **Tell your doctor right away if you develop any of the following symptoms while taking PROCYSBI:** headache, buzzing or “whooshing” sound in the ear, dizziness, nausea, double vision, blurry vision, loss of vision, pain behind the eye, or pain with eye movement.

The most common side-effects with PROCYSBI include:

- vomiting
- abdominal pain or discomfort
- headache
- nausea
- diarrhea
- loss of appetite or decreased appetite
- breath odor
- tiredness
- dizziness
- skin odor
- skin rash

Tell your doctor if you have any side-effect that bothers you or that does not go away.

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For more information, ask your doctor or pharmacist.

Call your doctor for medical advice about side-effects.

You may report side-effects to FDA at 1-800-FDA-1088.

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2013 Fall Research Grants Funded with Lay Abstracts



Robert Mak, MD, PhD, Principal Investigator Harold Hoffman, MD, Co-Principal Investigator

University of California, San Diego

“Role of Pro-inflammatory Cytokines and Inflammasome Signaling in Nephropathic Cystinosis”

\$259,754 – 2-year grant (February 1, 2014 – January 31, 2016)

Project Title: Role of pro-inflammatory cytokines and inflammasome signaling in nephropathic cystinosis

Objective/Rationale: The intralysosomal cystine accumulation in cystinosis leads to infantile nephropathic cystinosis (INC). Preliminary data suggest that cystine crystals activate the NLRP3 inflammasome and may be important for the pathogenesis of INC. We propose to characterize the expression of NLRP3 inflammasome components and downstream mediators in the mouse model of cystinosis (Ctns^{-/-}). The results of this investigation may pave the path for novel therapies for INC.

Project Description: We will characterize the time course (1, 4, 9 and 12-month of age) of expression of NLRP3 inflammasome components and downstream mediators and correlate with cystine content, renal dysfunction as well as perturbations in energy homeostasis and growth failure in the cystinosis mouse versus control mouse. We will evaluate the significance of pro-inflammatory cytokines and inflammasome signaling in INC via genetic and pharmacological approaches. We will compare the inflammatory and metabolic phenotypes of IL-6^{-/-}Ctns^{-/-} double knockout mouse, NLRP3^{-/-}Ctns^{-/-} double knockout mouse and ASC^{-/-}Ctns^{-/-} double KO mouse. We will also compare the

effects of anti-inflammatory therapy (IL-6 and IL-1 monoclonal antibody) on metabolic and renal function in cystinosis mouse.

Relevance to the Understanding and/or Treatment of Cystinosis: Pro-inflammatory cytokines may be important for the pathogenesis and complications of INC. While numerous cytokines have been implicated in this process, the specific mechanisms of inflammation are unclear. NLRP3 inflammasome responds to cellular stress and mediates the release of pro-inflammatory cytokines. This project is important as we will investigate the differential role of pro-inflammatory cytokines as well as NLRP3 inflammasome signaling in INC. Our hypothesis, if proven, will likely lead to novel therapy for INC.

Anticipated Outcome: By profiling the pro-inflammatory cytokines and NLRP3 inflammasome signaling in cystinosis mouse life cycle and correlate these perturbations with renal and metabolic phenotype, we expect to observe that NLRP3 inflammasome signaling cascade is more important than IL-6 cytokine in the pathogenesis of INS. By applying both genetic and pharmacological approaches, we expect to observe a significant improvement in renal and metabolic phenotype in cystinosis mouse.



Giusi Prencipe, PhD, Principal Investigator Fabrizio De Benedetti, MD, PhD, Co-Principal Investigator

Bambino Gesù Children's Hospital, Rome, Italy

“Inflammasomes Activation in the Pathogenesis of Cystinosis”

\$88,000 – 1-year grant (February 1, 2014 – January 31, 2015)

Project Title: Inflammasomes activation in the pathogenesis of cystinosis

Objective/Rationale: Cystine-crystal accumulation is the hallmark of nephropathic cystinosis. The role of these crystals in generating tissue damage is still unclear. We have recently demonstrated that cystine-crystals induce production of pro-inflammatory molecules, called IL-1 β and IL-18. This production involves activation of the inflammasome, a multiprotein complex responsible for the activation of inflammatory processes. Based on these data and on recent results, demonstrating that cysteamine bitartrate (Cystagon[®]) reduces oxidative stress in mouse models of chronic kidney disease, we hypothesized that the protective effects of cysteamine on kidney in cystinotic

patients might be explained not only by the ability to lower cystine levels, but also by a mechanism acting through the reduction of inflammatory processes.

Project Description: In this study, we propose to study *in vitro* the effect of cysteamine on inflammasome activation and investigate the potential mechanism involved in inflammasome inhibition. Moreover, we propose to evaluate in the Ctns^{-/-} mouse model whether the administration *in vivo* of cysteamine acts on inflammasome activation in kidney, bone, liver and muscle. Finally, we want to investigate whether treatments of Ctns^{-/-} mice with available inflammasome blockers affect inflammation in target tissues and the development of the disease.

Continued from previous page

Relevance to the Understanding and/or Treatment of

Cystinosis: This project will shed light on cystinosis pathogenesis, identifying the role of the inflammasome pathway, investigating an alternative mechanism of action of cysteamine in cystinosis and providing the rationale for novel therapeutic strategies that may complement conventional therapies used in presently available approaches.

Anticipated Outcome:

We expect to obtain further results supporting the hypothesis that inflammasome activation may play a key role in cystinosis disease, contributing to the development of the interstitial inflammation and fibrosis leading to end-stage renal disease observed in cystinotic patients.

Matias Simons, MD, Principal Investigator

Imagine Institute, Hospital Necker-Enfants Malades, Paris, France

“A *Drosophila* Approach to Nephropathic Cystinosis”

\$227,700 – 2-year grant (May 1, 2014 – April 30, 2016)

Project Title: A *Drosophila* approach for nephropathic cystinosis

Objective/Rationale: Lysosomes are commonly referred to as the sink of the cell. Our previous results, however, have shown that lysosomes actively control the uptake of proteins. This occurs via the so-called mTOR pathway, a major metabolic pathway, which is activated at the lysosomal surface. Activation occurs via growth factors and amino acids. Due to its amino acid transport function, we speculate that cystinosin contributes to mTOR signaling, and we would like to test this hypothesis by generating a fruit fly model for cystinosis. This model will also be used to screen for molecular targets in novel therapies.

Project Description: The aim of our project is to throw new light on cystinosin by studying its functions in the fruit fly *Drosophila*. Using *Drosophila* we could previously show that prolonged lysosomal stress reduces Megalin-mediated endocytosis on the apical surface of epithelial cells. The expression of the Megalin receptor was shown to be under the control of mTOR signaling. These findings were validated in the proximal tubular epithelium of the mouse, confirming the suitability of *Drosophila* for the study of lysosomal biology. We now want to test whether the absence of cystinosin affects lysosomal function and mTOR-dependent endocytosis in *Drosophila*. We will also use the *Drosophila* model to screen for molecular factors and test for food modifications that attenuate cystinosin phenotypes.

Relevance to the Understanding and/or Treatment of

Cystinosis: So far, most research on cystinosis has been performed on various cell types isolated from patients. One important drawback of this approach is the difficulty to establish adequate control cells. Mouse models, on the other hand, are expensive and time-consuming as well as difficult to study at the cellular level. Using a genetically tractable short-lived animal such as the fly offers several advantages, most notably the possibility to study cystinosin functions in native epithelial cells alongside with their unaffected control cells. Furthermore, fruit flies are famous for their screening power, which we will exploit to find suppressors of cystinosin phenotypes.

Anticipated Outcome: We expect to clarify a possible role of cystinosin in apical endocytosis as well as mTOR signaling in *Drosophila*. We believe that the obtained results will be of high relevance for the understanding of proximal tubular cell function, which is the main affected tissue in nephropathic cystinosis. Another goal is to establish a bone fide cystinosis disease model that can be exploited for screening approaches. Thus, we expect not only to contribute to a better understanding of nephropathic cystinosis but also to introduce novel molecular targets for treatment.

Fall Grants Funded: \$575,454 • 2013 Total Grants Funded: \$2,153,048

Published Studies by CRF-Funded Researchers (since our Fall 2013 issue)



“rAA V9 Combined with Renal Vein Injection is Optimal for Kidney-Targeted Gene Delivery: Conclusion of a Comparative Study” – Published May 2014 issue of *Gene Therapy* by Stéphanie Cherqui, PhD, University of California, San Diego.

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The Proximal Tubule in Cystinosis: “Fight or Flight?” – Published in the *Journal of the American Society of Nephrology* 2014 by Robert Chevalier, MD, University of Virginia.



“The Fate of Nephrons in Congenital and Heritable Renal Disorders” – Published in the *Journal of Pediatric and Neonatal Individualized Medicine* 2013, by Robert Chevalier, MD, University of Virginia.

“Responses of Proximal Tubular Cells to Injury in Congenital Renal Disease: Fight or Flight” – Published August 2013 in *Pediatric Nephrology* by Robert Chevalier, MD, David Harrison Professor of Pediatrics, University of Virginia, Charlottesville, Virginia.



<<<
“Time Course of Pathogenic and Adaptation Mechanisms in Cystinotic Mouse Kidneys” – Published February 2014 in *JASN* by Pierre Courtoy, MD, PhD, and Héloïse Gaide Chevronnay, PhD, de Duve Institute and Université Catholique de Louvain, Brussels, Belgium.

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“Dedifferentiation and Aberrations of the Endolysosomal Compartment Characterize the Early Stage of Nephropathic Cystinosis” – Published December 2013 in *Human Molecular Genetics* by Olivier Devuyst, MD, PhD, Division of Nephrology, Université Catholique de Louvain, Brussels, Belgium.



<<<
“Upregulation of the Rab27a-Dependent Trafficking and Secretory Mechanisms Improves Lysosomal Transport, Alleviates Endoplasmic Reticulum Stress, and Reduces Lysosome Overload in Cystinosis” – Published August 2013 in *Molecular Cell Biology* by Gennaro Napolitano, PhD, Sergio Catz, PhD, Scripps Research Institute, La Jolla, California.

CRF Science Report and Research Grant Updates

CRF research grant progress reports are published in the *Research* section on our website: 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

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2014 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 \$25.8 million, with every dollar raised going directly to cystinosis research.

The CRF will announce its fall 2014 call for research proposals and fellowships in September. The guidelines for applications will be available on the website.

Visit www.cystinosisresearch.org/research/for-researchers for details.

The Cystinosis Research Foundation utilizes a Scientific Review Board (SRB) comprised of leading international experts in the field of cystinosis. (List at 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 (CRF) 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. 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 446 registrants in 39 countries around the world. The site contains a Professional Research Portal for researchers who register to access and view de-identified, aggregate cystinosis patient information. (www.cystinosisregistry.org)

CRF is excited about the future of cystinosis research, and is grateful to its researchers for their interest in the cystinosis community and its children. We look forward to working together to find better treatments and a cure for cystinosis.



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

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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.

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.

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MISSION

The mission of the Cystinosis Research Foundation is to find better treatments and a cure for cystinosis by supporting bench, clinical and translational research. Since 2003, CRF has raised \$25.8 million for cystinosis research in an effort to find a cure.

EDUCATION

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

CYSTINOSIS RESEARCH FOUNDATION

The Wish that Changed the World.



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

In **2003**
CRF was Founded

To have
my disease
go away forever



DURING THE FOLLOWING

11 YEARS

AND WITH SUPPORT FROM ITS MANY FRIENDS, CRF HAS RAISED

\$25.8 MILLION

TO BRING NATALIE'S WISH CLOSER TO REALITY



RENOWNED **SCIENTISTS**
FROM **AROUND THE GLOBE**
attended the 2014
International Cystinosis
Research Symposium



Your generosity
has funded

1111
STUDIES IN
COUNTRIES



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)**



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

100
PERCENT

OF EVERY DOLLAR
DONATED GOES DIRECTLY
TO CYSTINOSIS RESEARCH

YOUR COMMITMENT
HAS GIVEN

NEW HOPE

TO **500** CHILDREN AND YOUNG
ADULTS IN THE UNITED STATES
WITH CYSTINOSIS

AND **2,000** CYSTINOSIS PATIENTS
THROUGHOUT THE WORLD



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.**



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