# Cystinosis MAGAZINE

FALL 2011 FOR FRIENDS AND SUPPORTERS OF THE CYSTINOSIS RESEARCH FOUNDATION

## Research Takes Center Stage

The Searcher

A Week in the Life

Many Hands Make Light Work

A Day of Hope and So Much More

***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 insidious disease, but cystinosis remains incurable.

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

Federal funding for research on cystinosis and other rare diseases is virtually non-existent and most pharmaceutical companies remain uninterested because financial rewards are too small.

Yet, while there is only a small number of patients who suffer from any given "orphan" disease, knowledge gained by studying one disease often leads to advancements in other rare diseases and more prevalent and well-known disorders.

Cysteamine, currently the medicine used to treat cystinosis patients, is also in clinical trials as a possible treatment for Huntington's disease, Parkinson's disease and NASH (fatty liver disease), which affect millions of people worldwide.

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, the CRF is the largest provider of grants for cystinosis research in the world, funding more than 84 studies and fellowships in nine countries.

CRF has raised nearly $16 million, which it has granted or committed to cystinosis research studies around the world. CRF's efforts have changed the course of cystinosis research and given new energy to its investigators and scientists.

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

CYSTINOSIS
RESEARCH FOUNDATION

**For more information about the Cystinosis Research Foundation, call 949-223-7610 or visit www.cystinosisresearch.org.**

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*Cover photo by Lars Wanberg*

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


**Cystinosis Research Foundation website www.cystinosisresearch.org**

A NOTE FROM NATALIE

# *Dear Friends and Family:*

I just started my junior year at Georgetown University in Washington D.C. I am studying psychology and French, and I am enjoying my classes and learning a lot. I wanted to get more involved this year, so I joined a club and decided to interview for an internship position.

I was fortunate to get an internship with Senator Claire McCaskill from Missouri. I am thrilled to be working on Capitol Hill this semester. It is a dynamic environment and an exciting job. I am having a wonderful experience learning more about our government and the American political system.

Some of my duties include answering the phones and escorting visitors on tours. I have met some very nice people from all over the world, as well as interns from other universities in the area. In order to get to my job from school, I had to learn how to take the Metro and so far I haven't gotten lost!

I decided to live in a dorm apartment with other roommates this year, instead of a single dorm room. I have three roommates and I like all of them. I am happy to be living in an apartment and am lucky to have very nice roommates. Having someone around to talk to and hang out with makes a difference.

Soon after arriving at school, I had a health scare and needed to find a nephrologist in Georgetown. Fortunately, Dr. Grimm, my nephrologist at Stanford and my favorite doctor, made some calls and arranged for me to see a nephrologist on campus. I feel much better now and I know that I have a good team of doctors in Georgetown in case I need to be seen again. I am feeling magnificent now!

Although I am really busy this year with school and my internship, I am having a great year. I am very fortunate to be meeting so many new people. I truly enjoy everything I am doing.

**I look forward to seeing many of you at the next event in California. Thank you for your love and support for everyone with cystinosis.**

Love, Natalie

# *Dear Friends and Family:*

**As the year winds down and the holidays approach, we are reminded of what is most important in life: family, friends and community. We have so much to be thankful for this year and so many promising things on the horizon – FDA approval of the delayed-release medication, new stem cell research discoveries, closer family connections – all of these contribute to the great sense of optimism that permeates our reality.**

This issue of *Cystinosis Magazine* celebrates our science community. We recognize and appreciate those individuals who are working tirelessly on behalf of our community and who have dedicated their careers to improve not only the quality of life for those diagnosed with cystinosis but also to find a cure for our children.

We also celebrate Raptor Pharmaceutical Corp. and Sigma-Tau Pharmaceuticals, Inc., two companies that over the years have demonstrated their commitment to the cystinosis community by advancing new treatments. They are stellar champions on behalf of our community.

We are particularly pleased to introduce Dr. Jennifer Simpson from the University of California, Irvine. She and her colleague, Dr. James Jester, were recently awarded two CRF research grants to study novel treatments for corneal cystinosis.

Corneal cystinosis is a painful eye condition that affects individuals with cystinosis. Early diagnosis of cystinosis is often made by a slit-lamp exam at an ophthalmologist's office. Upon examination, thousands of tiny cystine crystals may be seen in children as young as 1 year. Corneal cystinosis causes photophobia (severe sensitivity to light) requiring most patients to wear sunglasses and hats – even indoors – beginning at a very young age.

As cystinosis patients age, their eyes are damaged by a build-up of cystine crystals on the cornea. The painful symptoms are often described as similar to having sand in one's eyes. Every blink is a painful irritant and a reminder that the disease is progressive. If left untreated, the crystals accumulate on the cornea, and some patients have corneal transplants while others go blind in their twenties.

There is a treatment for corneal cystinosis, but it is a rigorous regimen that requires hourly dosing of eye drops. Although we are thankful for an eye treatment, more needs to be done. CRF awarded grants to Dr. Simpson with the express goal of seeking novel treatments. Dr. Simpson, in collaboration with Dr. Stephanie Cherqui and others, is working on better treatments and a cure for corneal cystinosis.

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CRF is strategic in its approach to funding research. We prioritize translational research – the process whereby results in lab studies are turned into new therapies for patients. This "bench-to-bedside" approach ensures that promising novel treatments will find their way into clinical trials.

We are guided by the extraordinary members of the CRF Scientific Review Board who evaluate and analyze every research proposal we receive during our twice-a-year announcement for research and fellowship applications. We are thankful for Dr. Corinne Antignac's leadership as the chair of the CRF Scientific Review Board.

This issue of *Cystinosis Magazine* includes an update on Dr. Cherqui's continued research success with gene and stem cell therapy. Her research and corresponding publications have resulted in two NIH grants – an enormous accomplishment, especially in light of the fact that the success rate for NIH research grants is approximately 17.4 percent, the lowest rate ever recorded. Her two NIH grants have leveraged CRF dollars and propelled her work into the international spotlight.

*We recognize and appreciate those individuals who are working tirelessly on behalf of our community and who have dedicated their careers to improve not only the quality of life for those diagnosed with cystinosis but also to find a cure for our children.*

We are excited to report that our researchers are publishing articles in prestigious journals at a rapid pace as cystinosis research explodes exponentially. Our research teams are making new discoveries about cystinosis and its complications every day. Even more exhilarating is the reality that the discoveries made by cystinosis scientists and researchers are offering hope to patients with more prevalent diseases, such as Parkinson's disease, Huntington's disease and NASH, a fatty liver disease.

We know you'll be moved by our family stories and inspired by the efforts of individuals who have made a difference in their own unique and inspiring way for cystinosis research.

This holiday season is a time to reflect on all we have to be thankful for, to count our blessings and to renew our sense that great possibilities pave our tomorrows. There is no doubt that this is an exciting and promising time for CRF and the cystinosis community. We are closer than ever to improving the quality of life for those with cystinosis and to realizing a stem cell treatment for those diagnosed with cystinosis. With you by our side, we continue our march towards our quest for the cure.

We are thankful to each and every one of you for driving CRF to heights even grander than we could have ever imagined. Thank you for your financial support, commitment and dedication to our community.

With blessings from our family to yours,

***Nancy, Jeff, Alex and Natalie***

Fun in the Sun for the Entire Family

# DAY of HOPE

## Cystinosis Research Foundation Family Conference

**Thursday, April 19 – Saturday, April 21, 2012**

Balboa Bay Club, Newport Beach, California

**Learn, laugh, share and celebrate for three exciting days at the 2012 Cystinosis Research Foundation *Day of Hope* Family Conference and Natalie's Wish Event on the beach at the beautiful Balboa Bay Club in Newport Beach, California.**

- Top cystinosis researchers from around the world will share their progress on gene and stem cell therapies, novel eye research and neurological issues related to cystinosis.
- Raptor Pharmaceutical will provide an update on the RP103 delayed-release medication plus Sigma-Tau will discuss the eye drop progress with the FDA.
- Cystinosis families from around the world will meet, socialize and share their personal stories of life with cystinosis, as they participate in a weekend of hope and inspiration.

**All conference sessions, conference meals and the Natalie's Wish event on Saturday night are free for cystinosis families.**

CYSTINOSIS RESEARCH FOUNDATION

Learn

Laugh

Share

Celebrate

**For more information about the conference or the hotel, visit www.cystinosisresearch.org or contact: Nancy Stack at 949-223-7610 or nstack@cystinosisresearch.org**

# Eyes Offer a Vital Window to Cystinosis Effects and Treatment

*Dr. Jennifer Simpson, at the University of California, Irvine's Gavin Herbert Eye Institute, is doing research, including possible stem cell transplantation in the eyes, to discover treatments for corneal cystinosis.*

**Imagine trying to live with a constant irritant in your eyes that the medical literature describes as "needle-like." A substance that can make the simple act of blinking a trigger for sharp, intense pain. A pain that only yields to partial relief when special drops are applied every waking hour.**

For many cystinosis patients, there is no need to imagine such an ordeal; they live it every day. That's because a major complication of their rare and degenerative condition is the formation of cystine crystals that cling to their corneas. The crystals form and accumulate as the patients age, causing a high sensitivity to light, corneal erosion, keratopathies and, in some cases, blindness.

*"A unique part of this research is knowing that through collaboration we are able to accelerate understanding. We all know we are working on something important, and that's a wonderful feeling."* DR. SIMPSON

Now the Cystinosis Research Foundation (CRF) is working to change that dynamic. It's funding research at the University of California, Irvine's Gavin Herbert Eye Institute to support novel treatments for corneal cystinosis, including stem cell transplantation in the eyes.

This stem cell arm of the UC Irvine team's corneal research draws on the success of studies performed by Dr. Stephanie Cherqui at The Scripps Research Institute in La Jolla, California. Dr. Cherqui's mouse model has proved an effective tool in multiple cystinosis research projects, highlighting the collaborative spirit that has fueled progress toward better treatments and, it is hoped, a cure.

Dr. Cherqui's research has shown that stem cell therapy can all but stop the accumulation of cystine in the body's tissues, which causes cell death and eventually cripples vital organs, especially the kidneys.

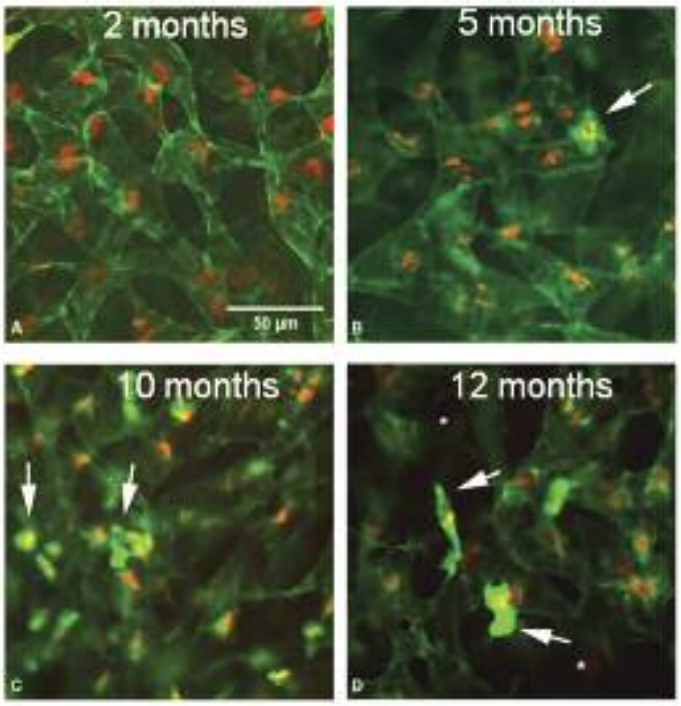
The eyes are also particularly vulnerable to the progressive effects of cystinosis. In the UC Irvine corneal research led by Principal Investigator Jennifer Simpson, MD, and Co-Investigator James Jester, PhD, stem cells are injected into the corneas of mice to see if the cells will transform into healthy, functional versions of those destroyed by cystinosis.

A second novel therapy the team is testing involves a rethinking of cysteamine eye drops, which are used to treat cystine crystals in the eyes. The idea is to add a very small chemical to the drops that acts as a vehicle for release of the drug over time.

The hope is that the drops would need to be administered only, say, once a day, as opposed to once an hour. Because the current protocol is such a burden and so impractical – the drops must be refrigerated – compliance is low, and the disease eventually takes its toll.

The investigators can't talk about the results of their research into these novel therapies because the findings haven't yet been published. But they do plan to present at the CRF-sponsored Third International Cystinosis Research Symposium on March 29–30, 2012, in Irvine.

What Dr. Simpson can talk about is the facet of her team's CRF-funded research dealing with the progression of cystinosis in patients. Using an elaborate microscope, the researchers can track the accumulation of crystals in the eyes of the mice without having to sacrifice the animals.



*Cystine crystals in ctns -/- mouse cornea.*

Because the eyes are easily accessed, examining them is often how cystinosis is first diagnosed in patients. What's more, as the patient ages, the amount of corneal crystal accumulation can indicate the course and severity of the disease itself.

"This can be a fast, inexpensive and effective way to screen as we evaluate the success of therapies," Dr. Simpson said. "What we're finding is that in mice the crystal volume, and thus the disease progression, correlates to what we've seen in humans.

"What's so important about this animal model is that it provides a quantitative, objective model for studying disease progression, which makes the opportunities offered by new therapies that much more exciting."

Dr. Simpson is just as excited about the chance to add her team's insights to the growing body of knowledge that sustains hope for cystinosis patients and provides motivation for researchers.


sigma-tau
PHARMACEUTICALS, INC.

**Karen Kuphal, PhD**
Sigma-Tau Pharmaceuticals, Inc.
Sr. Manager, Project Management

**Q Why did Sigma-Tau originally become interested in the eye drops?**

In 1995, National Institutes of Health (NIH) needed assistance in identifying a new supplier for the active pharmaceutical ingredient (API) being used in the cysteamine eye drop clinical trials. NIH had approached the National Organization for Rare Diseases (NORD) for their assistance in finding a partner, and in turn, NORD reached out to Sigma-Tau. Sigma-Tau helped find an API supplier and the relationship progressed. The original intention of both Sigma-Tau and NIH were for Sigma-Tau to take the lead on manufacturing and commercialization, while NIH focused on filing a New Drug Application (NDA). In 2004, the decision was made to transfer the regulatory responsibilities to Sigma-Tau for the filing of the NDA.

Cysteamine eye drops are exactly the type of project that Sigma-Tau is interested in. Cystinosis patients represent a small patient population with an extremely important unmet medical need. Sigma-Tau is enthusiastic about the partnership with NIH.

**Q Why does Sigma-Tau work with rare disease groups?**

It was the dream of our founder, Dr. Claudio Cavazza, to develop novel medicines to treat the unmet medical needs of patients with rare diseases. Sadly, Dr. Cavazza passed away only a few months ago, but his legacy continues. We work with patient groups because we feel responsible to support the patients and families that are affected by the diseases we treat. Also working with patients gives us tremendous insight into how medicines are used.

**Q What is the relationship with Sigma-Tau and the NIH?**

Our relationship with NIH has been ongoing since 1995. Dr. Gahl and the late Dr. Kaiser, both principal investigators of the eye drops, were key in getting Sigma-Tau involved in the project. We maintain strong relationships with the former Principal Investigator (Dr. Tsilou) and the recent transition to Dr. Bishop. Sigma-Tau is also exploring additional opportunities with NIH for the development of other therapies for orphan products.

**Q We understand that there was a manufacturing delay in the production of Cystaran®. Has the manufacturing delay been resolved?**

Cystaran® has two components in the manufacturing process. First, there is the API and second, there is the finished product. Currently, we have resolved the technical issue for the API. The issue surrounding the finished product is ongoing.

**Q When will Cystaran® be FDA approved?**

The complete resolution of the API and drug product chemistry and manufacturing control (CMC) information will be resubmitted to the FDA for their review (March 2012). If the review is satisfactory the drug may be approved.

**Q What stage of the FDA application process are you in?**

An NDA was filed in 2010. Due to a non-Good Manufacturing Practices (GMP) compliance issue with the third party manufacturing facility, FDA issued a complete response letter warranting CMC information to be re-filed to the FDA for review and final decision.

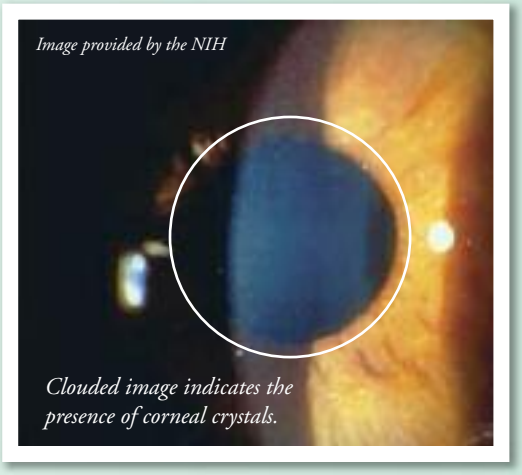


*Image provided by the NIH*

*Clouded image indicates the presence of corneal crystals.*

**Q Has Sigma-Tau filed the NDA?**

Yes we have filed an NDA.

**Q During the FDA approval process, can patients still get the eye drops from the NIH?**

The clinical trial is still open on an existing Investigational New Drug (IND) application in effect; and patients enrolled by NIH will receive cysteamine ophthalmic solution.

**Q Assuming the FDA approves Cystaran®, how will it be distributed? Will the drops be easily available, for example, will they be available at the local pharmacy?**

Currently we have plans in place for Cystaran® to be available through the same specialty pharmacy where cystinosis patients receive their Cystagon®. Upon placing an order for eye drops, the specialty pharmacy will ship them directly to the patient. At this time there are no plans in place to make Cystaran® available at the local pharmacy level.

**Q What is the protocol for administering Cystaran® – will they need to be taken every waking hour in order to be effective?**

Yes, that is the recommended dosage standard during the clinical trial.

**Q Will Cystaran® need to be refrigerated? Why does Cystaran® need to be refrigerated?**

The product is very unstable and easily oxidizes to cystamine if not stored frozen or kept refrigerated after opening. Oral cystamine doesn't break down the cysteamine crystals that accumulate in the corneas of cystinosis patients.

**Q Do you have any idea of what the cost will be for Cystaran®? Will it be covered by insurance?**

At this time there is no price structure in place. We have researched different scenarios and we are confident that insurance coverage for most patients will not be an issue. It should also be noted that Sigma-Tau has a very robust patient assistance and co-pay assistance programs which we plan to extend to Cystaran®.

**Q Does Sigma-Tau have any plans to improve the eye drop protocol by developing a slow/sustained-release eye drop?**

Yes, we are investigating the use of a slow-release gel, as a possible alternative to the current formulation.

**Q Sigma-Tau has dedicated resources to the cystinosis community for many years and has been a champion for the cystinosis community. Is there anything we can do as a community to help Sigma-Tau move the FDA approval process forward?**

Sigma-Tau values the support of the cystinosis community; however, at this time the principal obstacle to approval is a technical manufacturing issue that we are working through with FDA. We know this has been a long process, but in the end we know it is imperative to supply a GMP-compliant, FDA-approved product very soon. At this time we must meet the requirements of FDA and await their decision.

# The Searcher

## Dr. Stephanie Cherqui finds rewards and a range of emotions at the front lines of the fight for a cystinosis cure.

*By Dennis Arp*

Photo by Lars Wanberg

It is the ultimate researcher's dream to find a cure for a disease. It is a dream that Dr. Stephanie Cherqui has chased for 13 years, and it is now so close she can almost reach out and grab it. That makes this one of the most exciting and profound times in her professional life as a gene and cell biologist.

Dr. Cherqui and her team at The Scripps Research Institute in La Jolla, California, with the CRF Cystinosis Gene Therapy Consortium, which Dr. Cherqui chairs, are preparing to submit a clinical trial protocol to transplant healthy bone marrow stem cells as a possible treatment for cystinosis. It would be the first-ever clinical trial of stem cell and gene therapy targeting cystinosis.

In Dr. Cherqui's studies on laboratory mice, her research shows that bone marrow stem cells expressing a functional CTNS gene lead to significant decrease in cystine levels and cystine crystals in all tissues tested for the entire lifespan of the mice. The results harbor the hope for better treatment and ultimately for prevention of tissue injury in cystinosis patients.

Now that Dr. Donald B. Kohn and his research team at UCLA are hoping to launch the first clinical trial using human patients, "we should be able to find out if this is the way to go to develop a cure for cystinosis," said Dr. Cherqui, assistant professor, Department of Molecular and Experimental Medicine at The Scripps Research Institute.

"It's something we have been working hard to achieve, and now it's here, and that's very exciting. But this is a long process, and there are no guarantees. I feel responsible (for the patients in the study). That can be scary." DR. CHERQUI

"It's something we have been working hard to achieve, and now it's here, and that's very exciting. But this is a long process, and there are no guarantees. I feel responsible (for the patients in the study). That can be scary."

In moments such as these, Dr. Cherqui shows not just the depth of knowledge she brings to her research but also the wellspring of humanity. Thanks to events such as the CRF *Day of Hope* Family Conference, she has come to know the children and families directly affected by the life-altering complications of cystinosis.

*Continued on next page*

**To watch Dr. Cherqui's Day of Hope presentation go to:**
**http://www.youtube.com/watch?v=Es4Edx2PxGE**

Photo by Lars Wanberg


**Based on her CRF-funded research, Dr. Cherqui has recently had three articles published in prestigious scientific journals. The most recent was the January 2011 cover story, "Kidney preservation by bone marrow cell transplantation in hereditary nephropathy," in the journal *Kidney International.***

"I get to see them and talk with them, and I've really become attached to them," Dr. Cherqui said. "I know them as friends, and they know me and my kids. We're all part of this big group effort."

Often parents treat Dr. Cherqui like a hero – stopping to thank her for the research and the possibilities it holds.

"I say no, not yet," she related. "When we have a treatment for cystinosis – if we can confirm that this research is the right path to what we all want for cystinosis patients – then there will be an opportunity for us all to give thanks."

Knowing the patients' individual stories motivates Dr. Cherqui, she said. She has seen them struggle with day-to-day, quality-of-life issues such as being awakened in the middle of the night to take foul-tasting medication. She has seen their parents administer drops throughout the day to combat the pain of crystals forming in their eyes. She has talked with them about the effects of failing kidneys that keep them from leading normal, active lives.

"When I work, I can see their faces, and that pushes me forward," she said. "But that connection also has another side to it."

Dr. Cherqui knows that with any clinical trial, there is a risk. In this case, the participants will receive transplanted healthy bone marrow stem cells from a sibling in an effort to deliver the missing CTNS gene. Not only does she empathize with the patients in the study, she also doesn't want the results to disappoint her growing group of cystinosis friends.

"In mice, this therapy is really impressive," she said. "But there's always a question mark when you move to the next step. I'm the first to be impatient, but I know this process takes time and great care all along the way to clinical trial and, we hope, FDA approval."

That journey began in the late 1990s in her native France, where Dr. Cherqui worked as a graduate student in the Paris lab of Dr. Corinne Antignac. It was there that Dr. Cherqui launched her search for a cystinosis cure.

As a student, she quickly developed a deep interest in gene and stem cell therapy.

"Most of the time genetic disease affects children, and usually there is no cure, so it was the research area I wanted to work on," she said. "The only way to treat genetic disease is with gene therapy."

She met Dr. Antignac through her father, who was performing lab construction/remodeling. It was Dr. Antignac who suggested she target cystinosis. By taking on a rare, or orphan, disease, she felt her contributions could make a real difference in the lives of patients.

"To go from step one, which was finding the gene, to the final step, which would be finding a treatment – well, I don't know how many researchers get such an opportunity," Dr. Cherqui said.

Along the way, there have been countless days of diligence and, yes, many of tedious repetition in the lab. But there are also gleeful moments of revelation.

*Continued on next page*

## CRF Cystinosis Gene Therapy Consortium Members

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

**Stephanie Cherqui, PhD**
The Scripps Research Institute,
La Jolla, California

**William Gahl, MD, PhD**
National Institutes of Health,
Bethesda, Maryland

**Donald B. Kohn, MD**
University of California, Los Angeles

**Theodore B. Moore, MD**
Mattel Children's Hospital
University of California, Los Angeles

**Daniel R. Salomon, MD**
The Scripps Research Institute,
La Jolla, California

**Jerry Schneider, MD**
Dean for Academic Affairs Emeritus,
University of California, San Diego

**Nancy Stack**
Cystinosis Research Foundation,
Irvine, California

*Dr. Stephanie Cherqui with her husband Fabien Faucheux and her sons Noah (3 years old), and Matteo (6 years old).*

Photo by Lars Wanberg

*Frank Harrison has been Dr. Cherqui's research assistant for 5 years.*

*Brian Yeagy, PhD, is a post-doctoral fellow who joined Dr. Cherqui's lab 2 years ago.*

## Dr. Cherqui's Research Springs from CRF Support

Stem cell and gene therapy research projects led by Dr. Stephanie Cherqui are made possible by grants from the Cystinosis Research Foundation (CRF), which has supported her work since 2006. What's more, Dr. Cherqui credits the "preliminary trust and support of the CRF for her team," for receiving not one but 2 multiyear grants from the National Institutes of Health (NIH).

**"Dr. Cherqui's NIH grants are incredibly newsworthy. They further validate how highly regarded she is as a top researcher, because NIH grants are difficult to get."**

**NANCY STACK, CRF CO-FOUNDER AND CHAIR OF ITS BOARD OF TRUSTEES**

The latest NIH grant, announced in September, is a 2-year grant for "Kidney-targeted gene delivery for cystinosis." The long-term objective is to develop a kidney-specific gene therapy to introduce a functional CTNS gene using a viral vector injected via the renal vein of the patients to prevent or treat kidney defects in cystinosis patients.

"All of us at CRF are honored that our support has helped fuel the great progress that Stephanie and her team have achieved, and we're overjoyed that other organizations are now recognizing the importance of this research," said Nancy Stack.

In 1998, she was working with colleagues under Dr. Antignac's supervision at the French lab, searching for the mutation they could trace to the cystinosis gene, when one day a technician, Lionel Forestier, looked at his computer screen and said, "This seems like a real mutation."

The gene sequencing appears on the screen as a series of colors and peaks, but in this case where the technician expected to see red there was black instead. "And the peak was different," Dr. Cherqui added. "The base pair were different."

Researchers and technicians all looked at each other and smiled. "There was lots of excitement," Dr. Cherqui recalled.

Further testing confirmed: the team had discovered the cystinosis gene.

"It's a special feeling," Dr. Cherqui said. "It's like when I saw the effects of stem cell therapy for the first time."

In that instance, she took a kidney sample from a mouse and went to the microscope, where she saw that a lot of the matter cells appeared green. "That's weird," she thought at first. Then she realized it meant that the transplanted cells had migrated from the bone marrow to the kidney.

"I remember I spent hours at that microscope, taking lots of pictures," Dr. Cherqui said. "It's the kind of moment you live for. It makes all the work in the lab worth it."

One of the key ways her work becomes even more rewarding, she said, is through a spirit of collaboration. Since her days working with Dr. Antignac in Paris, Dr. Cherqui has readily shared insights, findings and even laboratory mice with researchers at rival institutions.

That isn't always the norm in the highly competitive world of biological and molecular research but Dr. Cherqui said she wouldn't have it any other way. "It's a small, supportive community," she said. "We're all trying to go faster."

Perhaps nowhere is that spirit of collaboration more tangible than at the International Cystinosis Research Symposium,

Photos by Lars Wanberg

*Frank and Brian work with Dr. Cherqui on stem cell research for cystinosis.*

organized and underwritten by the Cystinosis Research Foundation. The symposium brings together scientists and physicians from around the world to talk about the latest research towards better treatments and a cure for cystinosis.

Dr. Cherqui will help organize the third annual symposium March 29–30, 2012, at the Beckman Center of the National Academies in Irvine, California.

> "Dr. Antignac and Dr. Cherqui have put their egos aside to find a cure for cystinosis. By sharing the mouse model, Dr. Antignac saves each team up to 2 years of research, which is what it would take to create their own model." NANCY STACK

"Each time we meet, there's tremendous energy," she said. "We brainstorm ideas, some of which we can take directly to the lab."

Her role as symposium co-chair with Dr. Corinne Antignac, Dr. Julie Ingelfinger, and Dr. Elena Levtchenko, comes on top of her position as a member of the CRF Board of Trustees and its Scientific Review Board, as well as chair of the CRF Cystinosis Gene Therapy Consortium, which seeks to move promising therapies into human patients as soon as possible. She has also recently published articles in three prestigious scientific publications. Reflecting on her commitment to cystinosis patients, Stack seems to speak for the entire cystinosis community when she calls Dr. Cherqui "our rock star."

When Dr. Cherqui heard the characterization, she smiled, then gave an uncomfortable chuckle. After a long pause, she said, "That's really nice."

More silence.

"Let's wait and see," she said, finally.

## NIH Funding Grant

Dr. Stephanie Cherqui obtained a new grant from the National Institutes of Health (NIH), the nation's medical research agency, for her work on cystinosis. This new grant is a 2-year NIH Exploratory Developmental Research Grant Program supported by the National Institute of Diabetes and Digestive and Kidney diseases. This grant, entitled *Kidney-targeted gene delivery for cystinosis,* will support the optimization studies for renal vein injection of self-complementary adeno-associated virus (scAAV) expressing a functional *CTNS* gene to treat or prevent the renal defects in cystinosis.

Dr. Cherqui has received a previous NIH grant for 5 years to support the preclinical studies for the autologous transplantation of bone marrow stem cell genetically modified to express a functional *CTNS* gene.

For both these grants, the preliminary data necessary to obtain these governmental grants were generated with funding from the Cystinosis Research Foundation.

**NIH/NIDDK R21-DK090548-01A1**
**7/01/11–6/30/13 (2-year grant)**
*Kidney-targeted gene delivery for cystinosis*

To demonstrate that renal vein injection of self-complementary adeno-associated virus (scAAV) expressing a functional *CTNS* gene will treat or prevent the renal defects in cystinosis when delivered very early in the disease and ameliorate the renal disease if administered to older patients.

Amount: $236,875 per year

**NIH/NIDDK R01-DK090058-01**
**1/01/11–12/31/15 (5-year grant)**
*Lentiviral-transduced hematopoietic stem cell transplantation for cystinosis*

To develop a multisystemic strategy for the treatment of cystinosis and establish the pre-clinical studies for the transplantation of autologous hematopoietic stem cells genetically modified ex vivo to express a functional CTNS gene using a lentiviral vector.

Amount: $289,123 per year

# RP103: New Hope for Patients, Rare Opportunity for Company

*By Dennis Arp*

*Ted Daley, President, Raptor Pharmaceutical Corp.*

As Raptor Pharmaceutical Corp. plans its strategy to compete in an industry dominated by corporate giants, the company knows it has to pick its opportunities with great care and foresight.

Raptor officials say they couldn't be happier that they set their sights on – and put its resources behind – RP103, the delayed-release version of Cystagon® that offers new hope for cystinosis patients.

In July, the cystinosis community was equally excited to hear Raptor's announcement that key goals had been met by its pivotal phase 3 clinical trial of RP103. The study showed that the medication was able to control patients' cystinosis, as measured by WBC cystine levels, using a twice-daily dosing regimen vs. every six hours with the current standard of care.

The other important result is that trial participants experienced no unexpected serious safety concerns attributable to RP103.

The next step will be an extension study to gather more safety data as Raptor prepares to file for FDA approval early next year. Then, if all goes as hoped, approval could come as soon as mid to late 2012.

"This is definitely big news for the cystinosis community and for our company," says Raptor President Ted Daley. "It's wonderful to be able to give definitive news. The last few times we've reported on progress, we've said we have to wait for results. Now we have results."

The goal in developing RP103, or DR Cysteamine, is to greatly improve the treatment and quality of life for cystinosis patients. Dosing every 12 hours instead of every six will reduce stress on patients and family members, allowing them to lead more normal lives. For instance, those who participated in the study point to the great benefit of being able to sleep through the night rather than having to be awakened to take the next life-sustaining dose.

*Dr. Minnie Sarwal with Tina Flerchinger at the Stanford RP103 clinical trial.*

Because Raptor is FDA-regulated, company representatives have to be careful about making broad statements regarding RP103, Daley says. But those outside the company say the potential benefits are huge and hard to measure. For instance, some parents whose children are involved in the study say their youngsters are doing better in school because of the chance to sleep through the night.

Such tangible gains were envisioned by the Cystinosis Research Foundation (CRF) when it funded all of the studies that led to the discovery of the slow-release medication.

"We wouldn't be where we are in this process today without the CRF," Daley says. "Their support led to the proof of concept and this new formulation."

Once the slow-release discovery was made, a patent was issued at the University of California, San Diego. In December 2007, Raptor acquired exclusive worldwide licensing for the development of DR Cysteamine for cystinosis as well as for other potential indications, including NASH (fatty liver disease) and Huntington's disease.

For now, however, the focus is squarely on cystinosis, Daley says. Raptor is eager to begin making a difference in the lives of patients.

"Our first goal is to develop a product that targets a clear and meaningful unmet need," Daley notes. "That's true in this case, but the opportunity is especially appropriate

for our organization because of the expertise we're able to bring and to execute."

Raptor's diminutive size is a benefit here because its nimble, streamlined approach produces efficiencies larger companies would struggle to match. Because the population of patients is smaller than with some conditions, the company is able to build first name relationships with its ultimate customers – the patients, many of whom offer direct feedback to Raptor in helping point out areas of need.

"We can bring great focus," Daley says, "and we're thrilled that this process has moved as quickly as it has. We're all moving at full speed to get what everyone wants – an effective and safe medication for cystinosis patients."

"Raptor's second goal is to provide value beyond simply making the drug available," says Daley. "We are in a position to help people with cystinosis in a way that other, larger companies help their patients. We are currently in the information gathering stage, but now that we have positive phase 3 data, we will soon begin to execute on some of the ideas that we've been given at the recent cystinosis family conference meetings, and from our patient surveys. The whole company is excited." he adds.

"Everyone at our company is personally involved with the cystinosis community," Daley says. "We see first-hand the need and the opportunities.

"This isn't just a self-sustaining business. We get great fulfillment from seeing the effects of our efforts, and that's one of the reasons we intend to stay in the rare-disease segment."

*"We wouldn't be where we are in this process today without the CRF," Daley says. "Their support led to the proof of concept and this new formulation."* *Ted Daley*

raptor pharmaceutical corp.


*Families at the Stanford University RP103 clinical trial: seated Joey Jordan, Bailey DeDio, Patrick Partington, Tina Flerchinger and Jenna Partington; standing, Mary Jordan, (Joey's mom) Teresa Partington, (Patrick and Jenna's mom) Tim Boucher, (Bailey's grandfather) Kevin Partington, (Patrick and Jenna's dad) and Mark Flerchinger, (Tina's dad).*

# Cystinosis Research Holds the Potential to Help Millions Worldwide

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

STEPHANIE CHERQUI, PHD, THE SCRIPPS RESEARCH INSTITUTE, LA JOLLA, CALIFORNIA

**"Seven years ago CRF went out on a limb to fund a series of studies that would not have received funding from conventional sources. These studies resulted in the development of EC-cysteamine, and ultimately the Raptor Pharmaceutics RP103 formulation. These new formulations have helped make cysteamine easier to ingest and tolerate, and as a result have opened the door for treating other diseases with this, as yet, underused drug.**

**"A pilot study published earlier this year has shown that cysteamine may be a potential new therapy for the epidemic non-alcoholic liver disease (NASH). A multicenter Raptor Pharmaceutics study is being considered next year for the treatment of NASH."**

RANJAN DOHIL, MD, UNIVERSITY OF CALIFORNIA, SAN DIEGO

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

**"Currently it is estimated that 13–16 percent of all adults in the United States have chronic kidney disease (that is, with renal function less than 60 percent of predicted normal levels) and is associated with five-fold increased risk of premature death due to accelerated cardiovascular disease. New therapies are urgently needed for this large patient population."**

ALLISON EDDY, MD, SEATTLE CHILDREN'S HOSPITAL, SEATTLE, WASHINGTON

**"Corneal cystinosis offers several advantages as a model from which therapies for more prevalent eye diseases will also benefit. First, since the genetic defect has been identified, the disease mechanism is well understood. Second, there is now a well-characterized animal model that can be used to evaluate new therapies. Finally, while the number of affected patients is relatively small, cystinosis affected individuals and families are highly motivated and well organized, making them an excellent population for the orphan drug and device program at the U.S. Food and Drug Administration.**

**"Examples of such potential cross-over benefits include the use of stem cell transplantation and long-acting drug delivery systems. While both of these approaches are being developed for corneal cystinosis, they also have tremendous potential in other ophthalmic conditions. These include, corneal chemical burns, which results in corneal scarring; corneal surface dysfunction, which causes severe dry eye; and a number of conditions that require chronic drop therapy, most notably glaucoma and uveitis. With the well-defined clinical endpoint of reduced crystals, corneal cystinosis provides valuable scientific and regulatory validation for novel therapeutic approaches to these more common conditions."**

JENNIFER SIMPSON, MD, UNIVERSITY OF CALIFORNIA, IRVINE

**"We wouldn't be where we are in this process today without CRF. Their support led to the proof of concept and this new formulation (RP103)."**

TED DALEY, PRESIDENT, RAPTOR PHARMACEUTICAL CORP.

# CCIR Update

*Betty L. Cabrera, CCIR Curator in the lab at the University of California, San Diego.*

**Betty L. Cabrera, MPH, Curator of the Cure Cystinosis International Registry (CCIR), would like to share the successes achieved in the year since the registry's launch. If you or a family you know have not yet registered, don't miss this opportunity to be a part of this global movement to find a cure for cystinosis.**

**The Year in Review**

Since it's launch in August 2010, CCIR has collected vital research information from 289 people affected by cystinosis, reaching people in 30 different countries worldwide. As the number of registrants grows, CCIR becomes a more valuable tool for professionals who might have a stake in cystinosis; such as researchers who conduct scientific investigations, investors who invest in biotechnology, and clinicians who wish to know more about the disease. Thus far, close to forty professionals in these different fields have requested access to the anonymous information contained in the CCIR database. As intended, the registry is bringing cystinosis patients and their families within reach of the medical research community as never before thus improving the potential for breakthroughs in treatment and perhaps a cure.

CCIR CURE CYSTINOSIS INTERNATIONAL REGISTRY

*The CCIR allows us to focus for almost the first time on directions that are particularly relevant to the patients themselves.*

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

The representation of cystinosis patients in the registry still falls considerably short of the number of known diagnosed cases, however, we aim to increase registration in the year to come. Members of the CCIR Advisory Board are continually working to spread the word to family organizations and clinicians by reaching out to them individually and by representing CCIR at conferences in the U.S. and abroad. There has also been a major push to improve accessibility to the registry. The CCIR website is now available in Spanish, French and Portuguese, as well as English. In addition, the Assisted Registration Program has been implemented so that people who experience barriers to registration (such as limited literacy or computer access) can get help from their own medical providers.

We hope families and individuals who have already registered can also help us promote the registry to people they know. Be sure to mention that the benefits of registration lie not only in the distant future with potential medical breakthroughs, but immediate benefits can also be realized if registrants take advantage of the many features available on the website.

Among the most popular is the *Ask an Expert* feature that permits registrants to submit an anonymous inquiry to cystinosis experts. Answers to cystinosis-related questions are provided within a matter of days. Soon, a list of already-asked questions and the answers will be posted on the website for all registered members to see and learn from. The questions address some of the concerns of cystinosis patients, while the medical and research community can also learn from them. We encourage you to keep your questions coming!

**Register at www.cystinosisregistry.org**

*Continued on next page*

# CCIR: NUMBER OF REGISTRANTS AND IMPACT CONTINUE TO GROW *Continued from previous page*

**We thank all of the individuals and families who have registered and remind those who have not yet registered why CCIR is critical in the fight against cystinosis.**

### What is a patient registry and why is it important to the cystinosis community?

Many different resources and tools are necessary to make significant advances in medical research. Progress in rare diseases like cystinosis are often impeded by the lack of information available about the disease and limited access to volunteers eligible for clinical trials. Therefore, patients who are willing to provide information about how the disease has affected them and also make themselves available to participate in trials are among the most valuable resources we have.

A patient registry is a widely used tool to conveniently collect both data about a disease and information about potential clinical trial participants. A patient registry is any system that allows for the organized collection of data about disease outcomes in affected populations for a scientific, clinical or policy purpose. The CRF has aligned itself with cystinosis medical experts and organizations worldwide to create the first-ever international patient registry for cystinosis, Cure Cystinosis International Registry (CCIR). The express purpose of CCIR is to make information available to the research community and thus promote accelerated research in advanced treatments and ultimately to find a cure for cystinosis.

### How does CCIR work?

CCIR is a web-based application that relies on persons affected by cystinosis and their families to provide answers to survey questions carefully developed by a panel of expert physicians. The survey captures medical and family history of the disease, as well as clinically relevant information about how cystinosis affects various systems of the body and quality of life.

**Confidentiality and protection of privacy are top priorities so personally identifying information is protected using three layers of security.**

CCIR has a designated Curator to oversee the daily operations of the registry and to assist people with the registration process. The curator also acts as a gatekeeper of the profiles contained in the registry, ensuring that confidentiality is maintained.

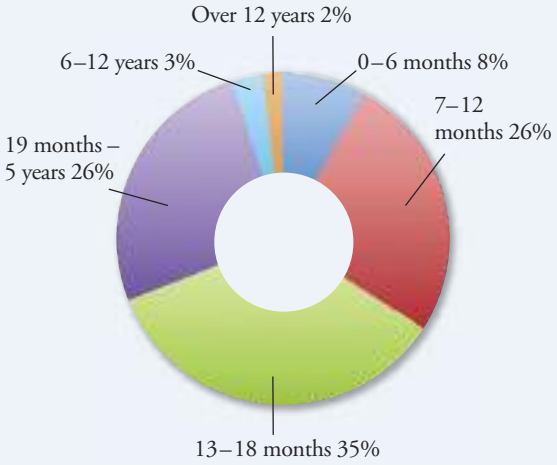
### What benefits can I expect to gain by registering?

When you register at CCIR, not only are uniting with hundreds of people from around the world to combat cystinosis, but you are gaining privileged and immediate access to a dynamic wealth of information about cystinosis never before collected in one place.

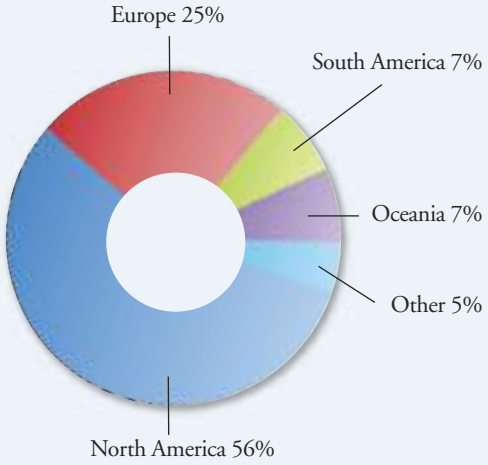
Upon completion of your profile, you will have access to the composite survey results and will be able see how the entire cystinosis community has responded to survey questions. You may be interested in checking the results frequently as more and more people register.

### Gender of Registrants


### Age at Diagnosis




### Geographic Location of Registrants



*The Cure Cystinosis International Registry allows cystinosis patients to submit their experience in privacy, while gaining access to anonymous information compiled from the entire group of registrants. Registered physicians can gain a wealth of practical clinical information about cystinosis based on the anonymous data as it accumulates.*

*Paul Goodyer, MD, Montreal Children's Hospital*
*Montreal, Quebec, Canada*

Through the *Ask an Expert* feature available exclusively to registrants, you will have the opportunity to submit questions about the disease for which you would like a cystinosis medical expert's response.

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

If you have any questions contact Betty Cabrera directly at curator@cystinosisregistry.org. Also, feel free to post a link to CCIR (http://cystinosisregistry.org) on your Facebook page.

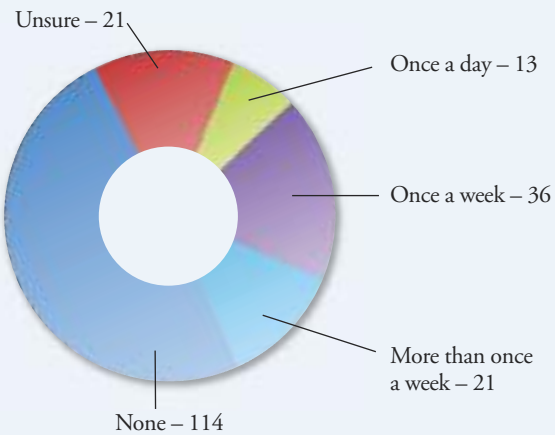
**Visit the CCIR website**
**http://cystinosisregistry.org**

## CCIR PARTNERS AND ADVOCATES

Cystinosis Foundation New Jersey Chapter
Cystinose France

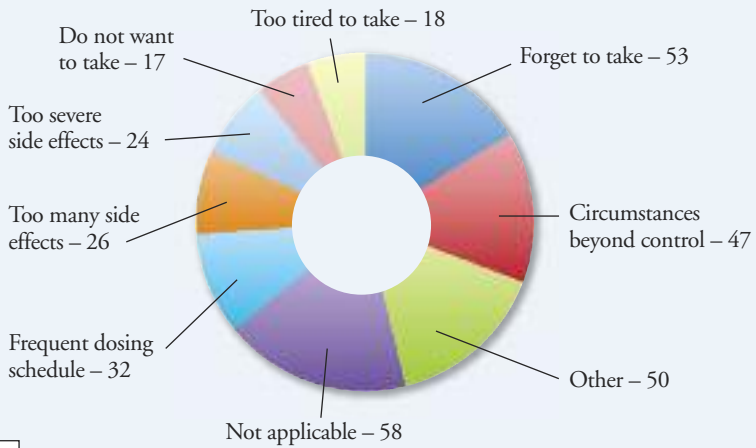
### Frequency Dose is Missed

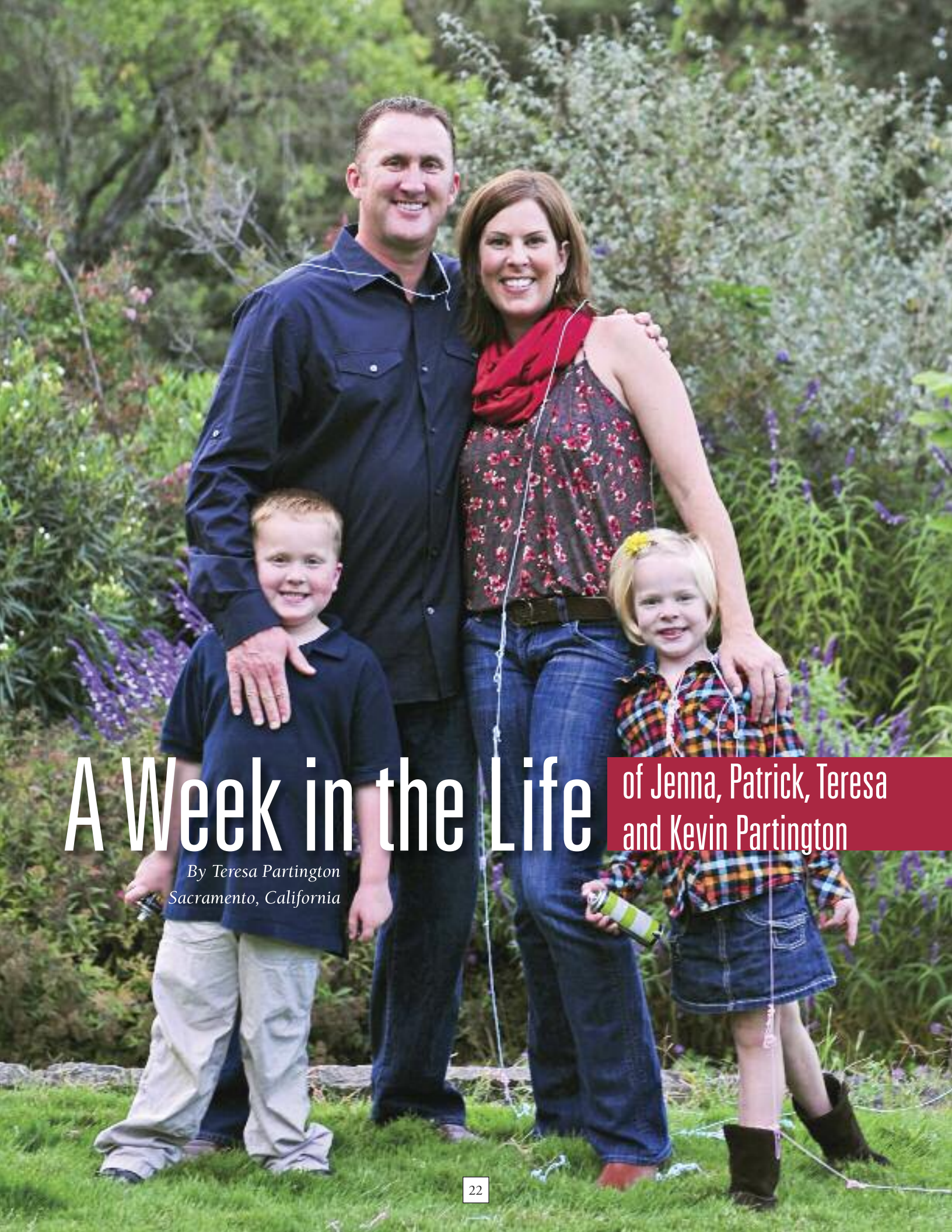
Number of Respondents




### Factors Contributing to Dose Skipping

Number of Respondents (Respondents could indicate more than one)





# A Week in the Life

## of Jenna, Patrick, Teresa and Kevin Partington

*By Teresa Partington*
*Sacramento, California*

## A Week of First Grade.

- Average pull-ups used overnight: 6
- Wet (soaked) medical-grade bed pads plus blankets and comforters brought to laundry from the night: 13
- Waters refilled in the night: 6 1-liter bottle refills
- Average times up per night: 1 (this is progress, people!)
- Vomiting: Patrick is the only one that vomits often: he averages 3 episodes of retching in 24 hours – all water, maybe meds
- Medications dispensed in 24 hours: 32 pills and 160 mls of 4 combined liquid meds.

### MONDAY, SEPTEMBER 29, 2011

**A restless night.**

We are trying to teach the kids to be independent in caring for themselves in the night; using the bathroom and changing the wet bed pads and blankets as needed. Jenna has done this just fine in the last week, but dug in her heels last night. I could have gotten out of bed to help her and all would have been settled in 60 seconds or less, but tough love had me digging my heels in, too – and it took about 15 minutes of Jenna yelling and banging on the walls for her to awaken Patrick, who ran to the bathroom for his standard midnight retching spell (sadly, that is so common now that I don't rise from bed for it, either).

Patrick proceeded to handle everything on his own: he threw up a few times, refilled his water, took wet bedding off and got himself fresh, dry blankets. Apparently, Jenna's ranting made him crazy too. In the hallway I heard a frustrated exhale from him (I could picture his eyes rolling), as he filled her water jug and took her a clean, dry blanket.

One could argue whether I should have gotten up to help or not…but I don't believe we are doing either of them any favors taking care of what they must manage on their own for the rest of their lives.

I was reminded as Patrick helped his sister of what a team they are. Their disease is so rare and so mysterious. That they have each other to relate to on this journey is a blessing.

Morning was smooth. We had eggs and hashbrowns for breakfast (they always crave a lot of starch and salt!) and we rode bikes to school. Today I will head to the pharmacy to pick up the months supply of medications.

### TUESDAY, SEPTEMBER 30, 2011

**Typical stats.**

Took photo of the laundry pile on my iphone.

Patrick threw up three times in 24 hours.

No soccer for P & J this year; the weather is still in the upper 90's and the kids literally "wilt" when we attempt recreation in the sun. I hate that they are missing this time with friends, and this opportunity to be a part of a team, however they are both participating in a class at our neighborhood gymnastics center which started tonight.

I had to run home mid-class to get Jenna a change of clothes, as bladder control has become a major issue.

*Continued on next page*

April Melarkey Photography

Who can blame the child who drinks two gallons of water per day? It is something that is going to need to be watched closely … since it's a newer problem.

## WEDNESDAY, OCTOBER 1, 2011

Patrick woke on his own for water and fresh bedding last night; and Jenna slept through the night. This is progress!

I forgot to give the kids their PolyctyraK (electrolyte) meds before we left for school and had to come home and get them to deliver it to the kids at school. I don't see how they would have enough energy for their day without a proper balance of electrolytes.

Monthly blood draws give us snapshots of what the kids' metabolic panel looks like, and it fluctuates greatly. We can help Jenna and Patrick's bodies function best by keeping them on a regular med schedule.

I am ordering hospital grade bed pads and diapers to create a care package for a family in the Midwest who have two children with cystinosis and very little money or resources to care for them. I wish I could buy a plane ticket and just go help out the family for a few days. I know how devastating it can be to have two sick children – to add financial stresses and lack of medical support in the area to that? It seems downright unbearable.

*Patrick's EKG at Stanford for the RP103 clinical trial.*

April Melarkey Photography

## THURSDAY, OCTOBER 2, 2011

Homework and school work are hard. Jenna, in particular, has trouble focusing on her school tasks. I can't help but wonder if it's a cystinosis thing, or if it's a normal kid thing. I worked for hours with the kids all week to study for a six-word spelling test – is this normal? Is this what it would be like if my kids weren't sick? Headaches, flat feet, core body strength, moles and birthmarks, coordination, attention span, bladder control, eating habits, night sweats, enlarged tonsils – these are just a few things that seem a bit "off" to me.

**Just what is cystinosis doing to Jenna's and Patrick's, little, young bodies?** From the day they were conceived something was wrong, and cystinosis has quietly and subtly been chipping away at their health and strength all along.

There are little rays of hope. Last Spring Jenna and Patrick were able to participate in an FDA drug trial for the new, slow release formulation of a life-preserving medication which is still scientifically titled "RP103."

We spent a good part of two months at Stanford University last April and May, where the kids were watched closely and received almost daily blood work to evaluate their white blood cell cystine count and general well-being. The kids tolerated it well and we made it a good, fun, family adventure. We traveled to the coast, toured Stanford University, visited the boardwalk at Santa Cruz and enjoyed the San Francisco Zoo.

What they missed in academics they made up for in life experiences (Jenna could run circles around the

hotel Laundromat) – though all of the time away from the classroom makes school work a struggle now. This new 12-hour time-release drug regime is a sign of progress, but it does nothing but buy us time. It is amazing and life-enhancing, but it is simply not enough. A timely, safe and effective cure is what Jenna and Patrick need to live the normal, healthy life that every kid should look forward to.

## FRIDAY, OCTOBER 3, 2011

My parents arrived today to watch the kids for the weekend – Kevin and I will travel with friends to San Diego to enjoy the wonderful Padres game package that we purchased at last year's Natalie's Wish event live auction!

I am coaching my mom on dispensing medications, handling the kids bedding and nighttime routine and how to handle an emergency. She and my dad have been a great help since the day the twins were born, and they know the drill. As always, my parents showed up prepared to enjoy their grandchildren, and very little sleep. After all, for the last 6 years the kids have been on a 6-hour medication schedule, which required a dose each night in the middle of the night.

## SUNDAY FOLLOW-UP

We arrived home from our San Diego trip to two very happy kids, and to their rested grandparents, who enjoyed completely uninterrupted sleep both nights they were here! My mom said, "This is unprecedented – this RP103 drug is the ticket!"

## MONDAY FOLLOW-UP

Friday's spelling test came back today and it wasn't pretty. My worries are ever-changing…and I'm now just as worried about the kids' cognitive health as I am their physical and internal health. Kevin says it's a marathon, not a sprint; I must remember this and take it all one day at a time.

*Jenna, Patrick and Tucker.*

April Mclarkey Photography

*CRF Board Member Denice Flerchinger, with daughter Tina and husband Mark at the 2011 Natalie's Wish event.*

# *A Powerful Rallying Cry*

*This is a transcript of a talk that Denice Flerchinger gave at this year's Natalie's Wish event on Saturday, May 21, 2011. Guests were mesmerized as a strong but nearly tearful Denice told how her daughter Tina faces life with the joy, excitement and anticipation of a healthy 8-year-old despite the unrelenting 24/7, 365 days medical regimen that is typical for cystinosis patients.*

Although most days I feel consumed by all that cystinosis has brought my family, tonight I feel JOY ... I feel HOPE ... I feel like a DIFFERENCE is being made!

Six years ago my daughter received a death sentence. Watching her endure one physical battle after another has been a painful experience. After one month of grueling tests in a hospital 320 miles away from home Tina was diagnosed with cystinosis. We were sent home with 33 doses of medication and told that our daughter would eventually succumb to a disease that would slowly destroy every organ in her body. There are no words to describe the grief and sorrow that come from being told "your child is terminally ill".

At that point in time, the only hope we had was getting through one more day. Nighttimes were long with all the medications given around the clock, the 24-hour vomiting, the all night feeds, and constant bedwetting. Tina still drinks two gallons of water every day. I remember administering her medications and crying at the same time, because the same medication that was extending her life was killing her.

**OUR ENTIRE FAMILY THANKS EACH OF YOU FOR LETTING US HAVE A LITTLE PIECE OF HEAVEN EVERY DAY WHEN WE LOOK INTO THIS LITTLE ANGEL'S EYES. WE KNOW ONE DAY TINA WILL BE CURED!**

That was then, this is now... FAST FORWARD to today. After years of hoping and praying a breakthrough finally came in the form of a new medication to replace the old unpalatable one. Tina got accepted into an FDA trial through Raptor Pharmaceutical in August. She made the required weight by one ounce. That was one of the happiest days of my life!

But it came with a cost – all of winter was spent traveling back and forth to Stanford Medical. It was one of the most difficult things we've had to experience – the long days of travel, being away from home, missing half the school year, not to mention the rigorous testing schedules and blood draws.

But it was so worth it! The new medication has freed Tina from a life of nausea and vomiting. She can go to school without taking meds, participate in recess, and is eating well. And the most exciting part – she can sleep through the night after six years. No more nighttime meds. That's exciting for mom and dad too!

The new medication was funded by the CRF. Each of YOU has made a difference and continues to in the lives of all those with cystinosis, as well as other diseases like Huntington's disease, Batten Disease, Parkinson's disease and NASH. I joined the CRF board in October 2010 and I am continually amazed with all that is happening in the way of new treatments and a cure. I know one day Tina will be thankful that you have helped her fulfill her dream of being a mommy.

Although Tina is thriving, the reality is her organs continue to slowly deteriorate with each passing day. She knows she is sick. She has shared with us that she only wants to be like other children. To date Tina has taken 46,895 syringes of liquid medication and 57,488 pills just to stay alive. Amazingly, Tina rarely, if ever, complains.

Recently Tina had surgery to close her gastronomy tube site. Her potassium got so low they called a Code Red and we nearly lost her. These children are so fragile. Their lives linger between life and death. We know God has something big in store for Tina. Her journey has only just begun!

Tina still has a multitude of health issues. Our latest challenge is dealing with Tina's eyes. As she grows the crystals in her eyes change shape and become like shards of glass. It then presents itself as a prism, reflecting light. This is very painful. She rubs her eyes and says it feels like she has sand in them. The only present treatment are eye drops that are hard to comply with – they must be administered every waking hour, remain refrigerated, and are not FDA approved, so they are hard to obtain and expensive. She will need to take the eye drops for the rest of her life or she will go blind. The CRF is funding research for new eye treatments and we are ecstatic about that.

Having a child with a terminal illness brings unimaginable heartache. It hasn't been easy, but your past, present and future support to cystinosis research gives us the hope we need to get through just one more day. None of this would be made possible without your commitment, the commitment of researchers, doctors and so many others who support the CRF.

It wasn't until we came into contact with the CRF that I felt hopeful in this terrible fight to save our daughter. I'll never forget Nancy's words, "I will go to my grave finding a cure for Tina, Natalie and all those with cystinosis." Believe me, she will but she needs our help!

Our entire family thanks each of you for letting us have a little piece of Heaven every day when we look into this little angel's eyes. We know one day Tina will be cured!

Photos by Bob Hodson Photography

*Corinne Antignac, MD, PhD; Ted Daley, President, Raptor Pharmaceutical Corp.; Stephanie Cherqui, PhD; and Ranjan Dohil, MD, who were all honored at the 2011 Natalie's Wish event.*

*Near the end of the evening the audience exploded with excitement when Hank Sturgis, Tina Flerchinger and Jenna Partington joined the sensational vocal group, Overtone, on stage as they all sang* When You Wish Upon a Star.

**To watch Denice's heartwarming talk go to: http://www.youtube.com/watch?v=9qQdjnpW04A**

# Day of Hope

...and so much more!

**Cystinosis Research Foundation Family Conference**

**Thursday, May 19 – Saturday, May 21, 2011**
Balboa Bay Club, Newport Beach, California

## Learn ☆ Laugh ☆ Share ☆ Celebrate

**The hopes of the children and their families are written in the stars.**

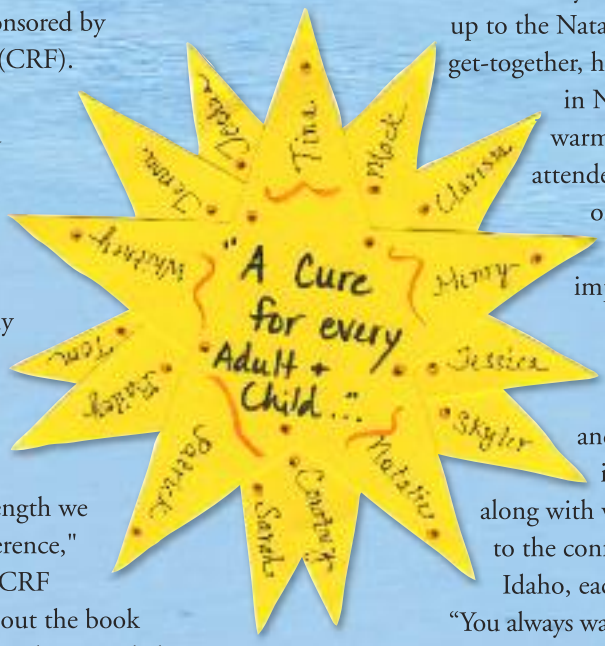
*"I wish I didn't have to miss so much school."*

*"I wish for Sam to grow up, live a healthy life and one day have a family of his own."*

*"May all the children reach their full potential and give back to the world with their own special talents."*

Inscribed on paper stars of gold, these inspirational messages and about 70 more helped launch the 2011 *Day of Hope* Family Conference sponsored by the Cystinosis Research Foundation (CRF). By sharing such wishful words for a brighter future, attendees introduced themselves to the other families as they also christened the feeling of camaraderie that would permeate the conference.

"We were so moved by the family introduction session that we copied all of the stars and put them into a book so that everyone who attended could remember the strong sense of community and the warmth and strength we found in each other during the conference," said Nancy Stack, co-founder of the CRF and chair of the Board of Trustees about the book of stars which was sent to the families who attended. (To view the stars, visit www.cystinosisresearch.org.)




The warmth carried throughout the conference, which started 3 years ago as a one-day meeting leading up to the Natalie's Wish event. Now it's a three-day get-together, held each year at the Balboa Bay Club in Newport Beach, California. And its warmth also carries throughout the year, attendees say, recharging the resolve of those on hand, strengthening them to take on the myriad of challenges that cystinosis imposes every day.

"We all face similar struggles, and to hear how others cope – the little pieces of wisdom that work in other families and might work for our family – that's invaluable," says Brian Sturgis, who along with wife Tricia and son Henry, traveled to the conference from their home in Sandpoint, Idaho, each of the past 3 years.

"You always want to know what's coming in the months ahead, and the families with older children are eager to help," Sturgis added. "Hank is just starting preschool,

so it's good to know how other kids have handled starting school. It's about the support we can give and the support we can get."

In addition to the time for cystinosis families to share concern, stories and useful information, the conference includes opportunities to interact with cystinosis researchers and doctors as well.

"Hearing the research progress reports is uplifting and inspirational," says Waterloo, Ontario, Canada resident Jody Strauss, who attended the 2011 conference with husband Trevor and daughters Gabbie (with cystinosis) and Chloe. "You get a real sense of their commitment to finding a cure.

"It's hard to describe how you feel when meeting and talking with them. They don't have children with cystinosis, but they work every day so our children can have a better life. So much progress is being made, and to hear about it first-hand is special."

For Kevin Partington, the three-day conference also provides validation that he and his wife, Teresa, can take back to Sacramento, where their Jenna & Patrick's Foundation of Hope has raised $1.5 million for research since their twin children were diagnosed with cystinosis in 2006.

"We can tell our donors about the ways their support is being put to good use," Kevin says. "I leave with a feeling of optimism I can share."

At the conference, Kevin makes a special effort to spend time with families who have a child that has been newly diagnosed with cystinosis. Those families are searching for optimism, he notes, as they experience what he calls the "Oh my God, what's going on?" phase.

"We can help them to understand that they don't have to be as scared as they might have been at diagnosis," he says.

Fear was trumping hope for Denice Flerchinger 5 years ago, when she and her family – husband Mark and daughters Tina, Catherine and Nichole – first heard of the Natalie's Wish fund-raising event that now culminates the *Day of Hope* conference.

"When Tina was first diagnosed in February 2005, I thought our whole world was coming to an end," says Denice, of Clarkston, Wash. "We cried for the first year. Now I just think what a blessing this has been for our family – that's how hopeful I am. We're changing history for a lot of families who will come after us."

Turning fear and sorrow into possibility and resolve was the idea behind the first conference 3 years ago, says

*Continued on next page*

*Nicole Cox and her husband Brandon listen pensively as they learn more about cystinosis and how to help their daughter Addison live and thrive with cystinosis.*

Photo by Lars Wanberg

# Day of Hope: Learn ☆ Laugh ☆ Share ☆ Celebrate

Nancy Stack. About 18 families attended that first *Day of Hope.* In 2011, 38 families shared in the experience.

"When people left the conference, we wanted them to share the experience with their family and friends. We wanted them to spread the word that discoveries about cystinosis are being made all of the time and that new treatments that will save our children are on the horizon," Nancy says.

For that excitement to build, Nancy knew it was important to break down any trepidation from the beginning. "Families had to feel comfortable enough with each other to communicate openly," she says.

That's where the notion of writing hopes and wishes on stars was born, linking families with the catalytic wish written on a napkin 8 years ago by Nancy and Jeff Stack's daughter Natalie – "to have my disease go away forever."

"I believe the stars set a tone," Nancy says. "Sharing the stars was a defining moment for the conference. When the researchers joined us in the room, the families felt safe and comfortable enough to share their stories and ask questions about research. By the time everyone left the conference, we felt connected in a deeply personal and profound way."

As the parents talked, their children played together in the care of counselors, building their own lasting connections. The most recent conference included a group sing-along on stage by the children, whose performance earned a standing ovation.

"It was a night to celebrate the cystinosis community," Nancy says.

Jody Strauss seems to speak for all the parents when she says of the entire *Day of Hope* conference, "It's an amazing experience." Then Jody offers one last gold-star wish.

"My hope is that every family affected by cystinosis would have a chance to attend this event."

*CRF Board Chair Nancy Stack and CRF Board Member Teresa Partington respond to questions from parents during the conference session.* Everything You've Always Wanted to Ask Other Parents.

*Paul Goodyer, MD, Professor of Pediatrics at the Montreal Children's Hospital in Canada, and member of the CCIR Board of Advisors, presenting the* History of Cystinosis *to conference attendees.*

*Playing a parachute game under the sun.*

**There are a number of Day of Hope videos, including several research updates at www.youtube.com/user/natalieswish**

# Day of Hope 2012: Thursday–Saturday, April 19–21

*Corinne Antignac, MD, PhD, Professor of Genetics at Hôpital Necker-Enfants Malades, Paris, France, and chair of the CRF Scientific Review Board, shares her research on the* Genetic Bases of Cystinosis.

*CRF Board Member Whitney Glaize (left), Tom Melang and Jessica Jondle answer questions about living with cystinosis as an adult.*

*Families gather to share information, exchange ideas and learn about promising new cystinosis research progress.*

*Children run and play at the* American Bar-B-Que Under the Stars *on Friday evening.*

*Time out from fun and games for a photo.*

*The Strauss Family enjoying a moment of reflection following a day of learning, sharing, laughter and hope.*

Photos by Lars Wanberg

# GABBIE STRAUSS WORKS AND PLAYS HARD WITH HER FAMILY AND FRIENDS TO FIND THE CURE.

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

*The food at Real Men Can Cook was great, and there was plenty of time to socialize with fellow friends of Gabbie Strauss.*

**If you are reading this magazine, then you probably know just how difficult it is for a family to manage a child with cystinosis. Cystinosis families have interrupted sleep, constant doctors appointments, pharmacy visits and we're always washing syringes, ordering meds, stocking up on diapers, washing sheets and the list goes on and on.**

Sometimes this life can get so busy, that when it comes to fundraising, we need a lot of help. We are so thankful that the Cystinosis Awareness and Research Effort received support when we really needed it. Through the help of our community and donations from families across Canada we raised more than $45,000 in 2011 and over $200,000 in 2 years.

Support from our community continued in 2011 with our first fundraiser of the year, *Real Men Can Cook*. Organized by staff from the City of Waterloo and employees of Waterloo Fire Rescue, this entertaining event featured over 70 would-be chefs from our community who each cooked a recipe of their choice in a competition for best appetizer, main dish and dessert. From ice wine marinated berries with maple syrup sabayon to pulled porked with caramelized onions, the 'real men' certainly cooked to impress celebrity judges *Looney Spoons* cookbook authors Janet and Greta Podlinski and Chefs on King, Head Chef, Peter Martin. Gabbie particularly enjoyed the cream puff swans and Uncle John's pulled pork and candied bacon. The event, which took place on April 2, 2011 and also featured live and silent auctions, a live band and dancing, raised $30,334!

Fundraising efforts continued on July 1 with the Street Impressions' 3rd annual Charity Car Show. Gabbie and her sister Chloe helped out with the BBQ and raised $2,200 for cystinosis research. Three days later, Gabbie was admitted to Sick Kids Hospital after a routine appointment revealed that she was acidotic, dehydrated and had low potassium levels. After four days of IV fluids, lots of hospital cafeteria bacon, movies and elevator rides, Gabbie was back to swimming, biking and enjoying summer once again. Hospital "sleepovers"

*Continued on next page*

*Gabbie with her dad Trevor, her mom Jody and her sister Chloe at the 2011 Day of Hope.*

Photo by Lars Wanberg


*Gabbie with her dad Trevor, her sister Chloe and her mom Jody.*

*Trevor, Jody and Gabbie Strauss with Indy race car driver, Mike Conway.*

*Gabbie promoting CRF with great enthusiasm!*

*Greg Marles, partner, Dundee Wealth Management, with Gabbie and Chloe at the golf fundraiser.*

*Gabbie and Chloe at Sick Kids Hospital in Toronto.*

are never much fun – neither are the tears and constant blood draws. We always pray for strength and endurance and like most cystinosis families, we somehow make it through. Those four days taught us once again to be thankful for times when we're not in the hospital and grateful to all the people who support cystinosis research.

More support for Gabbie poured in on August 17 at Dundee Wealth Management's annual golf fundraiser. Located in the small town of Listowel, Ontario, Listowel Golf and Country Club hosted this year's event, which attracted 144 golfers who swung, chipped and putted for cystinosis research. Gabbie handed out *Cystinosis Magazine* to the golfers, chased golf balls at the putting green competition and also showed off her own putting skills. The event raised more than $10,000.

As fall approaches, Gabbie will be starting junior kindergarten. Although she is very excited about starting school and learning new things, this new chapter brings additional challenges for us. We've been filling out forms and meeting with school staff to determine when and how to administer the medicines Gabbie needs during the school day. We hope and pray that this transition will go smoothly and that Gabbie will stay healthy and strong this school year.

We're so thankful for the Cystinosis Research Foundation, whose hard work and relentless determination has resulted in amazing progress for the cystinosis community.

We will continue looking for more fundraising opportunities and not stop until we've reached our goal – **to find a cure for cystinosis.** We want to thank CRF, everyone who has supported CRF and the doctors and researchers who have given cystinosis families hope for the future.

God bless you!

» *"We're so thankful for the Cystinosis Research Foundation, whose hard work and relentless determination has resulted in amazing progress for the cystinosis community."*

# S-A-M is Somebody Special

*By Ashton and Stephen Jenkins, Sam's mom and dad, Salt Lake City, Utah*

Sam's story is typical for children with cystinosis. He wasn't growing, he wasn't eating and he wasn't progressing. He threw up A LOT, soaked out of his diapers constantly and chugged water like a camel. We knew something was wrong. Our pediatrician knew something was wrong too, but he was at a loss as to what.

Our big break came when Stephen's aunt, Kathy Shaw, and grandmother saw Sam at a family reunion screaming for water. They thought his behavior and history bore an uncanny resemblance to that of Kathy's daughters, who were diagnosed with cystinosis two decades ago. We didn't think it possible or likely. Sure, Stephen might carry the defective gene, but what were the chances that Ashton did too? We asked for the test anyway, and were referred to a pediatric nephrologist. He was diagnosed a week after his first birthday.

The end of July marked the 1-year anniversary of Sam's diagnosis. While it has been a relief to finally have a diagnosis, it has brought with it the many adjustments familiar to all patients with cystinosis. Our daily routine keeps us conscious of the clock as we try to manage Sam's rigorous medicine schedule. In September of 2010, Sam got a gastric tube to help him get the nutrients he needs, as well as his medicine. He is currently on eight different medications and takes in over 1,200 calories a day. Since his g-tube was placed, he has gained 13 pounds, learned to walk, talk, and is a much happier little boy.

Despite being hooked to a feeding tube for a good portion of the day and dealing with frequent nausea, Sam finds a way to Rrrroar like a dinosaur, wrestle with cousins, and chase his bunny rabbits, Mabel and Matilda (also known as Bobble and Tata) around the backyard. He absolutely loves animals, especially lions and elephants. His favorite movies are *Fantastic Mr. Fox* and *Born to be Wild,* which he has seen in the theater five times now. We're currently preparing him for pre-school when he turns three next July. He knows how to spell S-A-M and is very happy with his accomplishment. He has an amazing attitude and strong spirit.

*Ashton, Sam and Stephen Jenkins.*

Sam has definitely transformed from a baby into a little boy. In the past month alone he ripped a curtain rod from out of the wall, popped his g-tube out of his tummy, smeared finger paints on our white couch, and threw his crayons in the dirty clothes hamper (discovered much too late). This is quite a change for us since he hardly left one spot for the first 18 months of his life. Each incident confirms to us that he can still be a normal little boy who loves all kinds of mischief and fun. We're so happy he finally has the energy to do so.

We are grateful for the hope that the Cystinosis Research Foundation has given to our family.

We know that Sam will be able to accomplish great things in his lifetime and that he will have a huge impact in the lives of others. We feel privileged to have such a wonderful little guy in our family and we love him very much.

# Gabe Stephenson: A VERY STRONG, VERY WISE YOUNG MAN

*By Tammy Stephenson, Gabe's mom, Dry Ridge, Kentucky*


Gabe's early years were much like any other cystinotic child – failure to thrive, vomiting, weight loss, no appetite. He was diagnosed at 13 months after being hospitalized for dehydration caused by a stomach virus. It was 1997 and research had just begun to blossom in the field of cystinosis. Cystagon was new at this time. The cysteamine eye drops were under research and Gabe was included in the study at the age of 3.

Since we believe in prayer and did not just want Gabe to survive, we began praying for Gabe to thrive and live a full life despite his cystinosis. Our faith in Christ began growing stronger every day. Many people were praying for Gabe, not just so we could cope with this dreaded disease, but that one day he would be cured.

Today, Gabe is a strong, healthy 15-year-old young man. Although he has not been cured of cystinosis, he has remarkable health. To look at him, you would never suspect he had any health issues. He is 5'4" and growing, and weighs 115 pounds without ever taking growth hormones.

His appetite is typical of a teenage boy – he's a bottomless pit! And, of course, the spicer the food, the better he likes it! Currently, the only medications he takes are RP103, potassium citrate, phosphorus, and cysteamine eye drops.

It is amazing to us how much research has already been accomplished in his lifetime. He has participated in all of the RP103 studies and is currently in the phase 4 extension study. Praise God! We must admit we do not miss the midnight doses!

Our family enjoys working out together at a local gym. Besides the desire for us to stay in shape, we wanted Gabe to build muscle in hopes that he would not have muscle wasting as he grows older. We first joined the gym when Gabe was almost 13. At that time, he was very "spindly" to say the least. Now his chest sticks out and he appears muscular.

> *"Just because you have a terminal illness, you do not have to live like you do."* GABE

Last fall, our trainer helped him prepare for a bench press competition. Gabe set a new Kentucky record for his age and weight bracket. At 14 years old, weighing in at 110 pounds, Gabe successfully bench pressed 115 pounds!

Besides lifting weights and trying to build muscle mass, Gabe enjoys running. In May, he completed his first 5K run in just over 26 minutes! Currently he is running 4 miles on a treadmill in just over 31 minutes!

He has already set higher goals for himself for both competitions next time!

Gabe's goals are all part of living that full life that we so often prayed for. Another reason he has these goals is that he wants to be a role model for the younger cystinotics. Last summer as we began phase 3 of the RP103 study in Atlanta, he enjoyed talking things over with the younger ones. He wanted to show them that they will be able to do the things their friends do. He stressed the importance of taking medications on a timely basis and exercising like you want to accomplish something. As Gabe says, "Just because you have a terminal illness, you do not have to live like you do."

We are currently making plans to attend the 2012 Natalie's Wish celebration. We hope to share more of Gabe's accomplishments with young cystinotics and to meet many of you there.

**In March 2011**, Vine Run Baptist Church's Youth Group sponsored a spaghetti luncheon in Gabe's honor. The church knows of his involvement with cystinosis research and offered this luncheon to show their support for Gabe and our family. We were able to tell guests what cystinosis is, how it impacts our lives, and where the research is heading. Approximately 50 people attended and donated more than $800 to support cystinosis research in Gabe's honor.

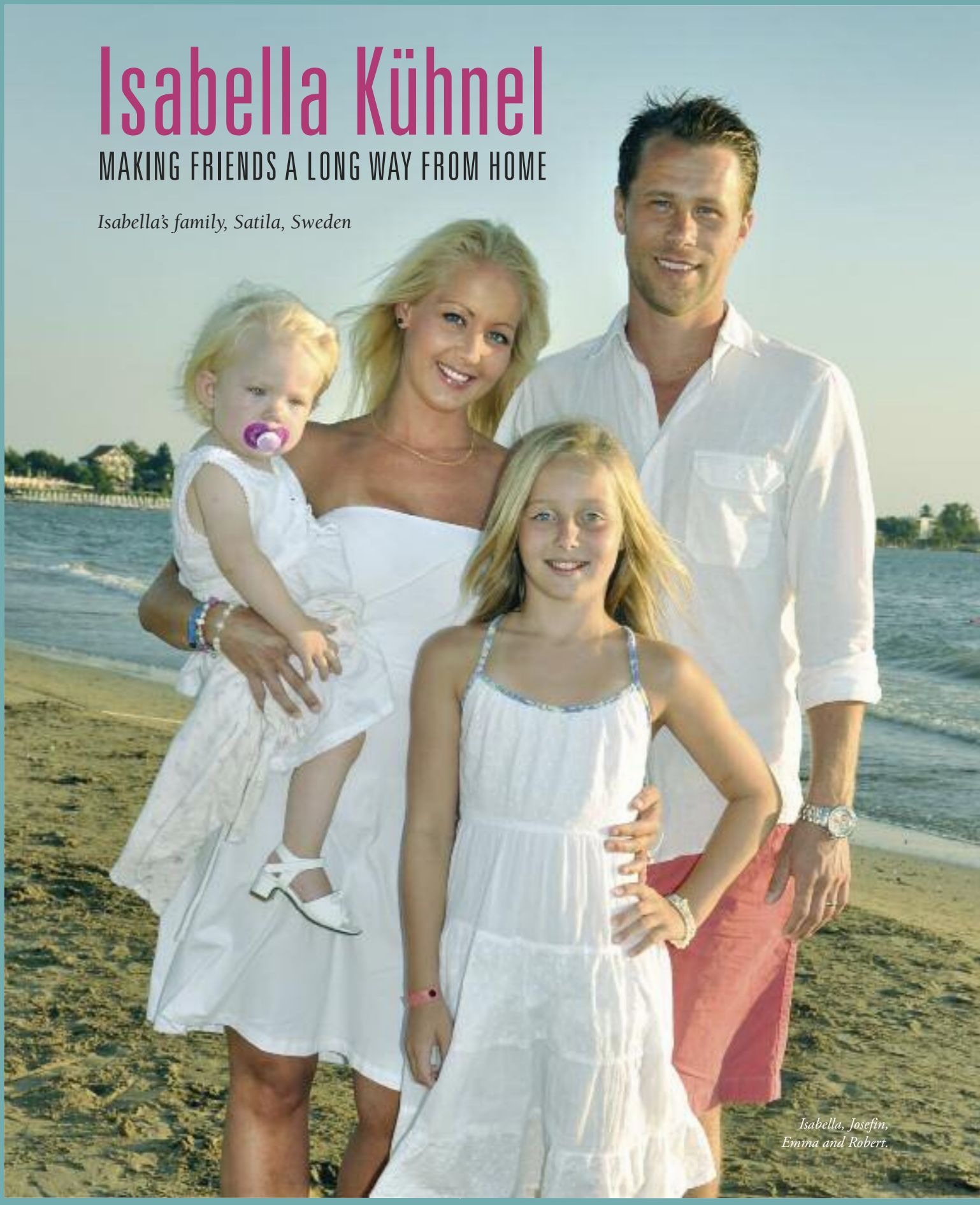
**In September 2010**, Gabe competed in a bench pressing competition, setting the Kentucky state record for 14-year-old males in his weight division. He weighed in at 110 pounds and successfully benched 115 pounds! His 2011 goal is 125 pounds. We'll let you know how he does!

**On May 7, 2011**, Gabe completed his first 5K run in 26:35 minutes. Next year, he plans to break this personal record. In training, he is running 4 miles on a treadmill in less than 35 minutes and plans to be at 5 miles in 35 minutes by spring. Gabe says it is important that he continues to meet these physical goals to encourage other cystinotics so they don't let the disease hold them back…Keep on running!

# Isabella Kühnel

## MAKING FRIENDS A LONG WAY FROM HOME

*Isabella's family, Satila, Sweden*

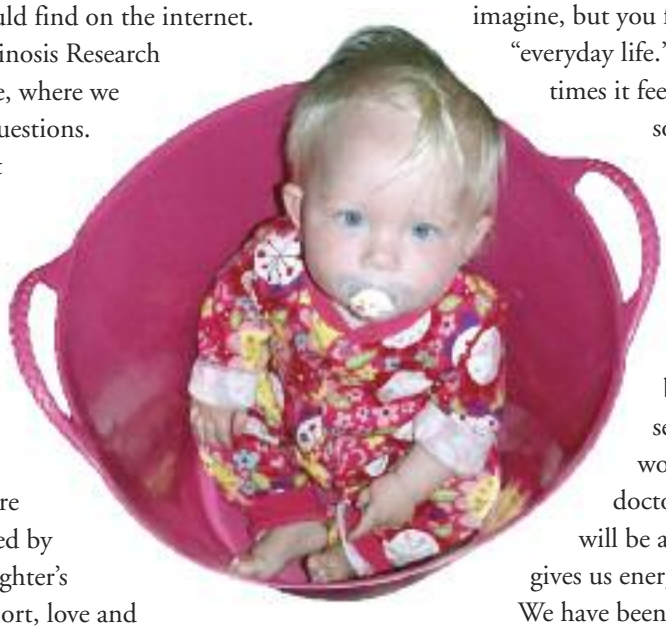


*Isabella, Josefin, Emma and Robert.*

# Isabella and her family find love, support and understanding at Day of Hope Conference.

Isabella was born in 2009 and seemed perfectly healthy until she was three months old. The symptoms started with vomiting, fever and loss of appetite. She lost a lot of weight and after three months of struggle – going between the hospital and home with various prescriptions of food and advice – she ended up in the emergency room. Two months and 13 doctors later, we got in touch with Dr. Sverker Hansson, at Queen Silvia Children's Hospital, Göteborg, Sweden, who had seen a few cystinosis patients before. Dr. Sverker is the most careful and loving person ever and he's given us so much support and help.

The fear, despair and lack of knowledge were unbearable. We read everything we could find on the internet. Finally, we found the Cystinosis Research Foundation (CRF) website, where we got some answers to our questions. Unfortunately, we had just missed the CRF's Annual *Day of Hope* Family Conference, but signed up for the 2011 family conference.



When the time finally came to attend the 2011 *Day of Hope* Family Conference, we were exhausted and overwhelmed by the uncertainty of our daughter's future. To receive the support, love and understanding of people we never met before was fantastic. If we hadn't gone to the conference, we probably wouldn't have made it through another day.

The days crossed into each other and getting a good night's sleep is a distant memory. The feeling of powerlessness makes it difficult to see any hope of improvement. Seeing our little girl go through things that you can only dream of in nightmares is terrible. And not being able to help her when she feels terrible makes you feel like a bad parent. The constant vomiting that forces you down on your knees to scrub floors, the endless medical preparations, cleaning, washing, stress, anxiety, headaches and fatigue – that's how we felt when we arrived at the conference in Newport Beach, California. *But how things changed in just a few days.*

There are only about seven cases of cystinosis in Sweden, so we didn't have many shoulders to cry on. At the *Day of Hope* conference we would come down for breakfast and know there were compassionate eyes who knew what kind of night we had been through. There were new friends to sit with if you needed to talk, something that is fairly normal for more common diseases.

The conference gave us hope, comfort and a glimpse of what the future holds for our children. Now, we hope, pray and believe that Isabella will be cured and that she will have a life like any other healthy young girl.

Life with cystinosis is something you can't begin to imagine, but you finally accept what we call the "everyday life." Sure, you look tired and sometimes it feels like it never ends (kind of true,) so what Robert and I do is relieve each other on the "tough" days. Robert and I started our own business the month Isabella got sick (before we knew about the cystinosis). It may not have made it easier, but practice makes perfect. We see this as a short period of hard work. And with the help of dedicated doctors and researchers we know there will be a cure in the near future, which gives us energy to continue.

We have been so inspired by the Stack family that we started a foundation in Isabella's name. We will try to get Sweden, or perhaps all of Scandinavia to pay greater attention to rare diseases and get more money for CRF researchers around the world. We are already contacting other families here (there are not many) who are also interested in going to next year's *Day of Hope* Family Conference.

We are truly blessed to live in Sweden with a healthcare system that provides us with anything we request or that our doctors order. It certainly makes life easier knowing that you do not have to worry about getting the best care for Isabella.

We look forward to seeing all of you again.

**Lots of love, Josefin, Robert, Emma and Isabella**

# The Flerchinger Family and Their Friends Do It Again!

*Tina (center) with her sister Nichole, mom Denice, dad Mark, and sister Catherine*

Photo by Lars Wänberg

## Never underestimate what a small community of family, friends and neighbors can do!

*By Denice Flerchinger, Clarkston, Washington*

On June 15 more than 300 people attended Tina's Hope for a Cure's 3rd Annual Wine, Stein & Dine, which raised over $90,000. The event took place at Roger's Toyota Scion Showroom in Lewiston, Idaho. Rich and Ryan Rogers graciously offered us their amazing venue.

Bright lights and live music set the mood for a perfect venue, as Tina mingled with the crowd. Guests were served international cuisine, Basalt Cellars wine, and Riverport Brewery beer. Auction items included a helicopter/jetboat trip on the Clearwater River, a Reba McEntire concert package, and a private cooking class for 12 at a local restaurant.

Jeff and Nancy Stack were the evening's "guests of honor." Nancy talked about the history of the Cystinosis Research Foundation (CRF), its current research, and how our cystinosis community is making a difference. She thanked everyone for their ongoing support, saying: "We are all in this together, and a cure is within reach." We are awed by Jeff and Nancy's unwavering kindness and generous support of our community.

*A crowd of 300 plus had a great time at the 3rd Annual Wine, Stein & Dine, raising more than $90,000 for cystinosis research.*

A documentary video, *A Day in the Life of Tina,* was shown, updating family and friends about the year Tina has had. There have been many challenges, but certainly the blessings have outweighed the challenges! There wasn't a dry eye in the room. After the video, Polly Blasko walked on stage and reminded the crowd that, "Together we are making a difference!" Fund-a-Cure followed and raised an astounding $33,000!

To say that we are humbled by the generous support of our community is an understatement.

Mark and I are both encouraged and optimistic by the CRF's role in the cystinosis community – funding research that will bring new treatments and a potential cure. The support of our community and so many other communities is helping the CRF continue to fund the much needed research that will bring a cure to Tina and so many others. We are thankful that we have had an opportunity to make this difference!

**To view A Day in the Life of Tina go to:**
**www.youtube.com/watch?v=VaSTUfctbLQ**

*Thank you to the lovely ladies of the board for working tirelessly to make this event possible. Holly Bonnalie, Mary Ausman and Sherry Seeh.*

# Par for the Course Not Good Enough!

**Geno Bonnalie recently proved that he's a heck of a good golfer, one tough warrior and a world-class inspiration.**

# 200 Holes of Golf in One Week

**Playing an amazing 2,000 holes of golf in one week – 16 rounds a day – Geno Bonnalie shattered the previous Guinness World Record by more than 150 holes, at Lewiston Golf and Country Club in Idaho.**

**By the end of his seventh day on Sunday, July 3, 2011 Geno looked like an emotionally and physically battered soldier who was returning home from a long, hard battle. With a crowd cheering him on as he finished his final round, Geno was in obvious pain. His right knee was stiff, his hands were blistered and his normally slender body looked gaunt.**

## Two More Records and a Few Near Misses

In addition to the 2,000 holes he played, Geno smashed two other records: Most Birdies in One Week at 491 (previously 250) and Most Birdies in 12 Hours at 67. If that wasn't enough, Geno also had seven eagles and a few holes-in-one that just barely failed to drop.

"It is really cool to say that I've played more holes – and made more birdies – in a week than anyone else on the planet, by a long ways," Bonnalie said.

## His Most Important Number: $15,000 to Find a Cure and Save a Life

But even with all of the record-breaking numbers floating around perhaps the most important one was the $15,000 Geno raised for ***Tina's Hope for a Cure,*** a partner of the Cystinosis Research Foundation.

Geno's numbers are staggering by any measure, but they pale when compared to those of his 8-year-old cousin, Tina Flerchinger, who was diagnosed with cystinosis when she was 17 months old.

## A Tough Young Warrior

Tina may be young, but she's no less a warrior, albeit an unexpected one. Since her treatment began 5 ½ years ago, Tina has taken nearly 60,000 pills and 50,000 syringes of liquid meds as she bravely battles the rare, genetic disease that is destroying every organ in her frail body. Unfortunately, unlike Geno's quest, Tina's battle goes on 24/7, 365 days a year until a cure is found for cystinosis.

Remembering his courageous young cousin, Bonnalie never considered quitting, but he admitted that "mornings were the hardest. " He would drag himself out of bed at 4 am, playing from 5 am until about 10 pm.

While Geno is the man who holds the new world records, he needed some help. "The maintenance staff and pro shop, I can't thank them enough," he said. "And all the members out here let me play through – I didn't wait a single time all week long. The second they would see me, they would pick up their clubs and run out of the way."

His wife, Holly reports that when he woke up Monday morning, Geno was upset. When she asked why, he said, "Because I gave up too quickly."

**Congratulations, Geno. In striking a blow – actually thousands of blows – to further the quest for a cure for cystinosis, you have shown us that one person can make a difference. And your remarkable accomplishment has inspired us all to look for ways in which we can make a contribution.**

for Cystinosis Research

Photo by Bob Hodson Photography

# Hank and His Friends Will Do Anything to Find the Cure

## 2011 Highlights in Honor of Hank Sturgis and the Cystinosis Research Foundation

*By Tricia and Brian Sturgis, Sandpoint, Idaho*

1

**Matt Gillis – Ran in the Portland Marathon for Hank – October 9, 2011 –** Matt Gillis calculated that it would take 58,000 steps to finish the Portland Marathon and asked his sponsors to donate $1 per step to help his friend Hank Sturgis. He finished in 03:38:03 and raised **$4,226** bringing his 2011 total to more than **$20,000**. Following the 24 Hour for Hank ski event, Matt found himself looking for new ways to make a difference in the lives of those with cystinosis. He felt there were more opportunities to raise funds and awareness for cystinosis than the yearly **24 Hours for Hank** events. He wanted to build a website that would become the central location for tracking events and accepting online donations, so he created ***Payin it Forward*** – www.payinitforward.com Visit the site to see what extreme event this exceptional young man is currently undertaking to support Hank and cystinosis research.

Photo by Lars Wanberg

2

**Fraternity SAE Sigma Alpha Epsilon –** SAE at Washington State University in Pullman, Washington, hosts an annual *Winter Olympics* with the Alpha Phi sorority. This is the second year in a row that they have chosen 24 Hours for Hank as their charity of choice. In 2010, they raised $1,400 and this year they have raised $2,000 for a total of **$3,400** for cystinosis in honor of Hank.

**"The generosity and compassion of our friends, neighbors and community is overwhelming and very much appreciated. We thank you for your support and commitment to Hank and to improving life for all those with cystinosis."** *Tricia and Brian Sturgis*

3 **Jennifer and James Riddle got married in June 2011, in Portland, Oregon**. The bride and groom asked their guests to donate to 24 Hours for Hank in lieu of wedding gifts. They have raised **$1,500** to date, and donations are still being received. We want to thank the happy couple for sharing their joy with Hank and the cystinosis community.

4 **Henry's grandparents, Dave and Mary Sturgis,** sponsored a BBQ for participants during a mountain bike event in July to benefit 24 Hours for Hank and raised **$265** from the BBQ dinner. Hank's Grandpa and Grandma Sturgis are passionate about volunteering and helping to raise money for CRF.

5 **Mexican Train Luncheon for Hank** – Pam Hausmann, Mary Sturgis, Debbie Johnson and Linda Currie organized an afternoon of pure entertainment playing the domino Mexican Train game while providing great food and libations for the participants. The suggested donation was $35 per person; they raised **$1,420** in honor of Hank.

6 **Katlynn Souers, (circled above) of Spokane, Washington,** is 13 years old and has been raising money for Hank since she was 9 years old. Her first fundraiser in 2008 was a cookie sale. In 2009 and 2010 she organized *Henry's Helpers* to do chores for neighbors then donating the money to 24 Hours for Hank. This year, she hosted 14 children for an afternoon of fun, games and crafts. She raised **$288** and is planning to host additional events with the goal of raising a total of $1,000 for cystinosis research.

7 **Lunch with Ben Stein, at Bottle Bay Restaurant on Lake Pend Oreille in Sandpoint, Idaho**. Lunch with Ben Stein was auctioned off at the 24 Hours of Schweitzer Ski Event and was purchased by Phil and Linda Currie, Edward and Rebecca Hawkins and Doug and Lodi Hawkins for **$3,000**. The luncheon event, held September 2, 2011, was enjoyed by all the lucky participants.

*Ben Stein, Doug and Lodi Hawkins, Phil and Linda Currie, Rebecca and Edward Hawkins*

To start your training for Hank's 2012 events go to: www.24hoursforhank.org


# HOLT MAY HAVE CYSTINOSIS BUT NOTHING HOLDS HIM OR TEAM GRIER BACK

*By Jason Grier, Holt's dad*
*Huntersville, North Carolina*

*Holt's sister Mary Logan, his mom Chrissy, Holt, his dad Jason and his brother Jack*


While living with cystinosis for 6 years has redefined our definition of "normal," we are and have been an extremely fortunate family and we work hard to never take that for granted. We continue to be blessed and humbled every day by the kindness, generosity and selflessness of our family, friends and the entire cystinosis community. There is simply no way that we could have made it as far as we have through this journey without all of your support – we continue to say thank you.

Holt had a great summer with several adventures. In addition to a family beach trip, "Mimi" and "Grammy" camps and a couple of visits to Carowinds theme park – Holt went to Atlanta to visit the Georgia Aquarium, The World of Coke and saw an Atlanta Braves baseball game. One of the Baltimore Orioles actually gave him a game ball! After a visit to the Great Wolf Lodge, a tour of the garage with his favorite NASCAR driver (Max Papis) and swimming every day in the backyard, it was on to football season. Holt cheers for the Davidson Day Patriots and the Lake Norman Giants. He is also a big Carolina Panthers fan (he wears his Cam Newton jersey to the games)!

While Holt still has all of the same symptoms and issues that you have read about many times before, he rarely (if ever) complains. His eyesight continues to deteriorate and his muscles are under-developed but his bravery and courage (medication every six hours) inspire Chrissy and me every day (We like to say: "We're TIRED but INSPIRED").

Holt loves to play with "race cars," play Wii and color and paint. He spends some part of every day running, laughing and playing with his brother Jack and sister Mary Logan. It is not uncommon to hear him giggle down the hall!

Having shared many times the trials and tribulations of living with cystinosis, Chrissy and I wanted to share with you a quick update on Holt and also some insight into how we keep a positive perspective on cystinosis and life in general.

Knowing how easy it is to let this disease and all that goes with it get you down, we continually find inspiration from those who have fought courageous battles. For us, it is through others that we garner our strength; we thought it might be helpful to share some of what inspires us in others facing similar challenges.

**What follows is one such example:**

In 1993, just weeks before he passed away after a courageous bout with cancer, former North Carolina State basketball coach Jim Valvano delivered one of the most riveting speeches the sports world has ever heard. I will never forget the day I heard it. The speech stands as one of the seminal moments in my life and it has been a beacon to which I return anytime Chrissy and I need a refresher course in courage and hope.

I have tried to capture some of Valvano's comments but the real value is seeing him yourself. Fortunately the speech is available on YouTube. I think you'll agree that watching it was time well spent.

**From Jim Valvano's ESPY Speech**

When people ask me how I get through life or each day, it's the same thing. To me, there are three things we all should do every day. Number one is laugh. Number two is think. Spend some time every day in thought. Number three is, have your emotions moved to tears, could be tears of sadness or tears of joy. Think about it. If you laugh, you think, and if you cry, you think. That's a full day. That's a heck of a day. Do that seven days a week, and you're going to have something very special.

*Mary Logan, Holt and Jack*

# Together We Are Making a Difference

**Joshua's Journey of Hope** worked for months organizing its first online fundraising auction that took place September 14, through September 23 raising over **$10,000** for cystinosis research.

*Joshua Clarke*

More than 250 items were donated from merchants and venues throughout California offering significant discounts of up to 30 percent. The auction provided bargains on theme park tickets, gift certificates to restaurants and golf courses, wine tasting tours, special hotel packages and exotic vacation getaways.

Because of the success of the first JJOH auction, plans are already underway to offer even more exciting items for the next year's auction. For more details, visit **www.joshuasjourneyofhope.org.**

Joshua's Journey of Hope

**Harbor Day School Sixth Graders Make a Difference for Natalie's Wish** Last May over a conversation on AIM, Will O'Connor mentioned to his classmate and friend Carson Shea, that he was attending the Cystinosis Research Foundation fundraiser that evening. Carson wanted to know what cystinosis was and why he was attending. After Will explained that his cousin Natalie had the disease, Carson wanted to know more about cystinosis.


*Will O'Connor, 12 years old*

After their online chat, Carson took matters into her own hands. She contacted her Harbor Day School classmates and told them about Natalie and how cystinosis affected her life. Carson wanted to help Natalie and all of the kids with cystinosis so she asked her friends to bring a donation to school the next day. "Any amount will make a difference," she told them.

The next day, Will was surprised when so many classmates walked up to him and gave him money for "his cousin's disease." When he got home and emptied his pockets there were bills and coins totalling **$300.**

Will was moved by this act of kindness shown to him by his classmates, especially Carson Shea.

CYSTINOSIS RESEARCH FOUNDATION
NATALIE'S WISH

**Landon Cystinosis Awareness Bracelets**
Cathy, from Cathy's Creation, learned about Landon and cystinosis through Landon's mom, Lauren Hartz. Cathy created a beautiful high-quality bracelet to raise awareness and support cystinosis research. The bracelets, named for Landon, are available at **www.cathyscreationsjewelry.com** for $40 and are made of sterling silver ball beads, blue crystals and a blue ribbon awareness charm. With the holidays approaching consider one of these beautiful bracelets for someone special and join us in the quest for the cure!

# Many Hands – Large and Small – Make Light Work

Jessica DeDio, Bailey's mom, decided to raise money for cystinosis research by bringing candy bars and cookies to the office and offering them for sale. Next month, Bailey's suggestion is to provide Slim Jims, chips and candy. Her office mates at Acralight Skylights loved the idea of raising money for Bailey and to date have generated **$865** to help find a cure for cystinosis. Thank you, Jessica!

*Bailey DeDio likes to have fun when he's not thinking of ways to raise money for CRF.*

**Tim Madden Ran the Western States 100 Trail Endurance Race for Jenna & Patrick's Foundation of Hope (JPFH)** – Tim Madden, a neighbor and good friend of the Partington family, ran in the endurance race to raise money and awareness for JPFH. The run, which took place on June 25 and 26 started at the base of Squaw Valley in Lake Tahoe and ended in the city of Auburn.

Tim completed his 100 mile run in less than 30 hours achieving his personal goal and raised over **$10,000** for the JPFH. Jenna and Patrick, along with Tim's friends, enthusiastically ran the final leg of the race to cheer him on to the finish.

*Chaz Sheya, Kevin Partington, Pete DeMello, JR Riddell with Jenna and Patrick*

## Lemonade Stand for Tina Flerchinger and Cystinosis Research

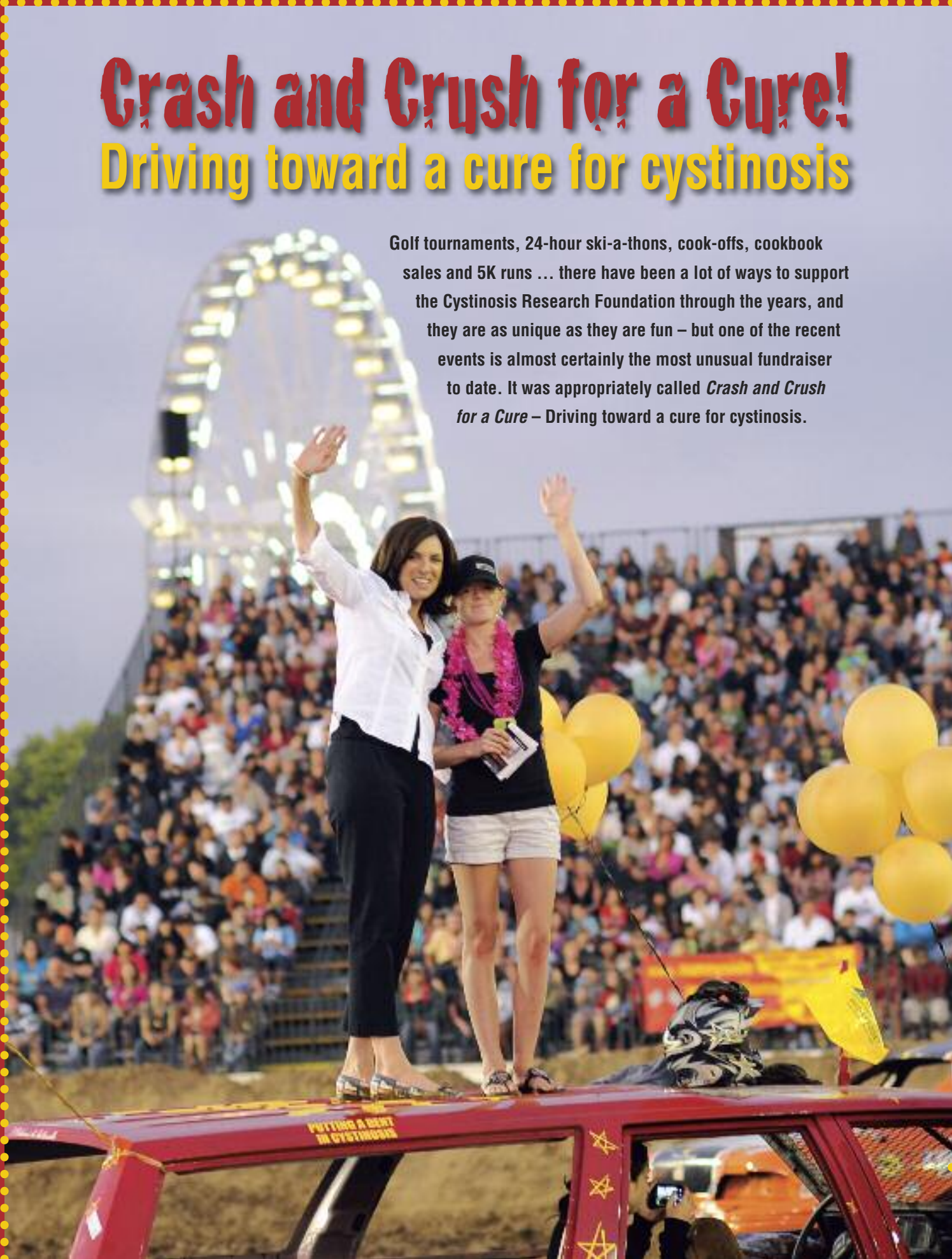
Gabby Trigsted is a schoolmate and good friend of Tina Flerchinger. Gabby and her older sister Emily wanted to support Tina by attending the Wine, Stein and Dine fundraising event this past June, but when their mother Wendy explained the event was for adults, not children, the girls declared they would host their own fundraising event and opened a lemonade stand.

They raised $89 and proudly presented their earnings to Denice, Tina's mom, exclaiming 'this is for Tina's cure."

# Crash and Crush for a Cure!

## Driving toward a cure for cystinosis

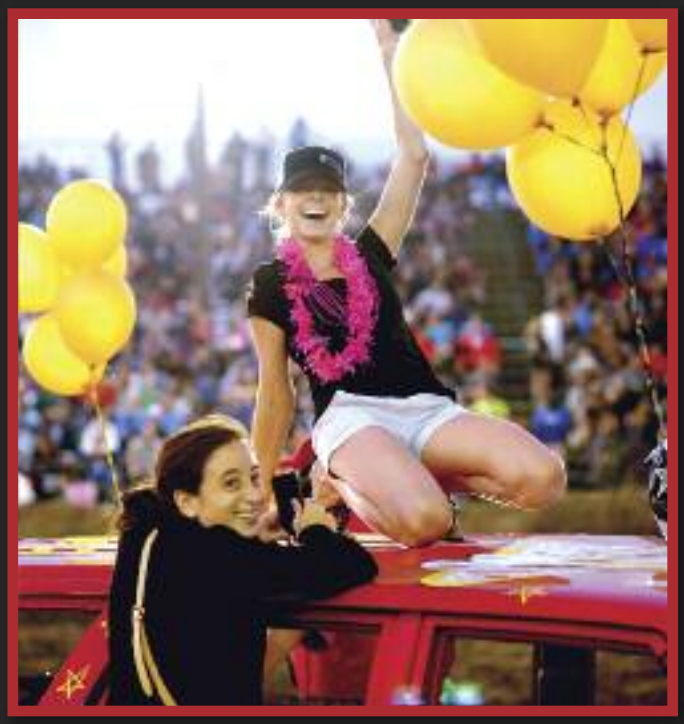
Golf tournaments, 24-hour ski-a-thons, cook-offs, cookbook sales and 5K runs … there have been a lot of ways to support the Cystinosis Research Foundation through the years, and they are as unique as they are fun – but one of the recent events is almost certainly the most unusual fundraiser to date. It was appropriately called *Crash and Crush for a Cure* – Driving toward a cure for cystinosis.



*Natalie with Billy Skeffington, president of Ben's Asphalt of Orange County who made the event possible.*

Ben's Asphalt of Orange County donated a car to Natalie's Wish to be auctioned off at the Orange Crush Demolition Derby at the Orange County Fair on Saturday night, August 13.

Ben's decorated and prepared a 1984 Buick Station Wagon especially for Natalie, using her favorite number, 21 (she turns 21 in February) and painting the car her favorite color, red.



*Natalie Stack and Christina Dialynas party with a sold-out crowd of more than 9,000 attendees, before the real fun begins.*

A sold-out crowd cheered as 19 derby cars took to the field for an unbelievable 45 minutes of crashes and crushes until the last car moving was declared the winner. Unfortunately, it wasn't Natalie's car, but there is always next year!

We want to express our sincere appreciation to all those who were part of the design and construction team at Ben's, putting in countless hours on the car and organizing the event. Special thanks to Billy Skeffington, John Skeffington, Teri Braden, Bill Grond, Megan Daniel, Joe Zumwalt, Doug Klevies, Eric Bajza, painter Ron Redman and Eric of the Sign Shop was responsible for the special car graphics.

We are also grateful to David Carchidi, of APT Companies, for interviewing Natalie and Nancy and recording the event on video.

Visit our YouTube channel to experience the evening's fun and excitement.


To watch the fun go to: www.youtube.com/watch?v=WQfTs56_Ual

# Even Eating is Hard

## When You Have Cystinosis

*By Whitney Glaize, Orlando, Florida*

**We are proud to launch *Living with Cystinosis,* a column we hope will help families cope with some of the daily struggles they face with cystinosis. Our first article is written by Whitney Glaize and deals with a struggle that many families have with their children – feeding and eating issues. Whitney's personal experience and professional training allow her unique insight into the everyday challenges that so many families face.**

**Whitney Glaize has lived with cystinosis for 32 years. She received her master's degree from the University of Central Florida in 2003 and her bachelor's degree for the University of South Florida in 2001. She is a speech language pathologist in Orlando, Florida, working primarily with young children.**

### MY STORY

Eating cessation is one of the early symptoms of cystinosis, so it is not unusual that as a young child with cystinosis, I struggled with feeding and eating issues. Like most children diagnosed with infantile nephropathic cystinosis, I stopped eating at about 15 months old. I ate normally until shortly before diagnosis, so my parents were surprised and puzzled when I suddenly refused to eat solid foods, even those I had once willingly eaten. As I grew sicker, I lost all interest in food and became obsessed with water.

At 22 months my electrolytes and cystine level were under control but I could not overcome my eating/feeding issues. I had a severe food aversion that remained for years. I did not eat solid food willingly or normally until I was 15 years old.

My cystinosis diagnosis came years before g-tubes were used, so my parents were never given the option to tube feed me. Fortunately, I loved whole milk and survived my early years by drinking a gallon of it each day. My parents also had success getting me to drink some juices.

The turning point came around age 15 when I got my first job, working as a waitress. For the first time in my life I was interested in food – how it looked, how it smelled and how it tasted. Until then, I had survived on baby food, chocolate milk, chocolate pudding, orange juice, grape juice, Hawaiian Punch, hotdogs and baked potatoes. It was a miracle given my limited diet that I consumed enough calories each day to grow and look healthy.

I think my parents would agree that it was a constant battle and one of the most difficult challenges of coping with cystinosis. My pediatrician had told my mom that a child's brain grows to 80 percent of its size by the time he or she is 3 years old. Consequently, my mom calculated every calorie and every ounce of food I ate because she was afraid I would suffer brain damage and be mentally disabled.

As a child I had terrible gag reflexes. The sight of a tongue depressor would make me throw up; I could not wear turtle neck sweaters until age 6; and I could not wear a birthday party hat because the elastic band around my chin would make me gag. My gag reflex and my insatiable thirst made it nearly impossible to get any solid food down. I didn't want to eat; I just wanted to drink water and nothing else. And drink I would … until I threw up.

As a speech language pathologist I work with many children who have feeding and eating issues. Many of them have a feeding tube of some kind. To help them, I have attended many workshops and seminars.

I believe there are many reasons why children with cystinosis don't eat. The medications taste horrible and have unpleasant gastrointestinal side effects. I took the original form of cysteamine, which many children and adults were unable to tolerate. I'm sure I associated the taste of the medicine with food. I also think that there are behavioral

reasons why children with cystinosis don't eat. For me, it became a battle of wills – a control issue. I was constantly being told what to do, what to put in my mouth, what not to do, when to get blood drawn, when to take medicine, etc.

When I was 8 years old, I had a swallow study done at NIH. They found no physiological reasons for my swallowing problems. There was some mild reflux (a side effect from the medicine) but nothing that would explain why I wouldn't eat.

Dehydration almost certainly played a role in my aversion to food. There is such a strong desire to drink that food becomes unimportant. The medications and electrolyte imbalance that caused vomiting became a regular part of my day. The fear of gagging and throwing up, further reduced my willingness to eat solid food.

As I got older and more medically stable, my feeding skills were delayed. When I decided I wanted to eat, I couldn't. I didn't know how to chew, how to coordinate swallowing with tongue movements, etc. Since I didn't know how to eat, I didn't.

Finally, while working in the restaurant, and as the social pressure and my desire to fit in with peers increased, I decided to overcome my fears and learn how to eat.

If you sat down to dinner with me today you would never know that for the first 15 years of my life I didn't eat. I have a normal diet and I'm not at all picky. I eat everything and enjoy eating just as much as the next person.

## MY RECOMMENDATIONS

**Here are some suggestions both from my personal experience and professional training that may help families to encourage their children to eat:**

- Never force feed your child. Research shows that forcing a child to eat can be traumatic for a child and do emotional harm.
- Try offering juices and liquids. Children with cystinosis have unquenchable thirst, so drinking is highly desirable.
- Try mixing water with new drinks or juices. Because they are obsessed with water, children with cystinosis may try a new juice if it is diluted by water.
- Don't mix medicine with food because it could make children think that all food tastes like medicine. Also, Cystagon® and other supplements taste so bad they can't be hidden in food.
- Let your child touch, smell and even play with food, even if they don't eat it.
- There may be such a thing as too much therapy or counseling for eating issues. I believe my family overused these approaches and I became even more averse to food. Years later, after the counseling and feeding therapy had concluded, I chose to eat.
- If possible don't put your child on a continuous feed/drip at night or during the day. Use bolus feeds to develop a sense of hunger.
- Try speech therapy/occupational therapy to desensitize your child to textures.
- When I was a child, oral motor therapy didn't exist and I would have benefitted from it.
- Introduce food in small steps: look at it, smell it, touch it, put it on your child's lips, touch it to their tongue, put a very small amount in their mouth, and lastly swallow a small amount. This procedure may take several weeks and if at any point the child gags or throws up, stop and start over at a later time. Never expect a child to take a bite of something new and swallow it. Baby steps.
- Use "food chaining-mixing." Combine tolerated food with a food that is not tolerated. For example, if your child loves ketchup, try mixing ketchup with mashed potatoes and slowly decrease the amount of ketchup added to the potatoes.
- Learning to chew was a challenge for me. There are oral motor exercises to strengthen the jaw and increase jaw mobility.
- Remember that your child will eat when he or she wants to, not when you want them to. If there is too much emphasis on it, it is possible, perhaps even likely, that eating will become a battle.
- **Be patient. It will take time.**

# Love for Landon

*By Lauren Hartz, Landon's mom*
*Pittsburgh, Pennsylvania*

*Lauren and Jimmy Hartz with their son Landon.*

Landon was born on April 8, 2010. He weighed 7 pounds, 4 ounces and was just perfect. He ate well and gained weight at a normal rate. Around seven months we noticed that he wasn't gaining weight at the same rate as he had before. From this point on, his weight gain was less and less until he didn't gain weight at all.

At 10 months, his pediatrician referred us to a gastroenterologist. Over the next four months, Landon had visits with the gastroenterologist, multiple tests and scans, sessions with a dietician and then a feeding team. The vomiting began at 13 months and the gastroenterologist ordered a scope. We were sure that it was acid reflux. Landon had the scope done but there were no signs of acid reflux or damage to the esophagus.

We were relieved but upset because we still had no answers. The doctor decided to admit him to Children's Hospital in Pittsburgh for a feeding evaluation.

We received instructions for admission five days later. The doctor collected a urine sample during the scope, which indicated that there was sugar in his urine. We met one of the hospital's nephrologists who talked to us about Fanconi Syndrome. They knew that was a possibility because they found substances in Landon's urine that should not have been exiting his body. The nephrologist explained that Fanconi Syndrome is sometimes a result of another metabolic disorder called cystinosis.

On June 8, 2011 Landon was diagnosed with cystinosis. We had prepared ourselves for the news but it still wasn't easy to hear.

Landon was in the hospital for 25 days. He endured multiple trips to have a NJ tube placed and then two surgeries to have a GJ tube placed and then a g-tube. He experienced countless blood draws, being disrupted in the middle of the night for vitals and to get his weight, having IVs put in and pulled out (sometimes by nurses and sometimes by Landon!). But, our little fighter never ceased to amaze us. He was quite a popular little guy and stole the hearts of many nurses, doctors and staff.

Now, almost two months after discharge from the hospital Landon is thriving. He is receiving bolus feeds every three hours during the day and a continuous feed throughout the night. He now weighs over 17 pounds!

He takes six medications throughout the day: Cystagon® every six hours, Prevacid twice a day, Polycitra, Enrythromycin and Levocarnitine three times a day, and sodium phosphate four times a day. He experienced some vomiting, diarrhea and constipation but his belly issues seem to be improving. He is in physical therapy and will begin occupational therapy to work on feeding issues.

Our hope for Landon is still the same as it was before he was diagnosed. We want him to be happy and to be the best Landon that he can be. We hope that he will not just sit back but will face cystinosis head on, and that he will educate himself and others.

Finally, we hope there is nothing, including cystinosis, that will stop him from accomplishing his goals or preventing his dreams from coming true.

We have made it our job to raise awareness and contribute to the research. We owe it to everyone who is affected by this disease, and especially to our baby boy!

**The Cystinosis Research Foundation's** primary focus is a cure for this rare, metabolic disease. In April 2009, scientists from Scripps Institute in La Jolla launched a 5-year path to clinical trials in children with cystinosis. The trials will test new gene therapies believed to hold the first real promise of a cure.

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the May/June 2011 issue of
*Multihousing Professional* magazine


# Come together right now

Natalie Stack, 20, began her Junior year at Georgetown University this year. She's bright, beautiful, and clearly the pride and joy of her parents, Nancy and Jeff. She also happens to have cytinosis, a rare disease that will challenge her until there is a cure.

From the minute you meet Natalie, you know she is meant for greatness. She exudes poise and strength, well beyond her years. And she is determined.

Jeff Stack, managing director of Sares-Regis, is a multifamily owner, manager and developer in Irvine, Calif., and long-standing pillar of the industry. He and his wife, Nancy, have launched a private seige on this rare disease for which there is yet a cure. But the future looks bright.

On the eve of her 12th birthday in 2003, Natalie made a birthday wish that no child should ever have to make.

After leaving a doctor's appointment, Natalie and her mom stopped to have lunch at a local restaurant.

While discussing plans for her birthday party, Nancy asked her daughter if she had thought about making a birthday wish. Natalie said she did not have a wish because she wasn't sure what type of wish she should make. She then asked if her wish had to be one that could come true. Natalie then picked up a napkin and scribbled something on it. She folded the napkin and put it in her pocket.

As they were walking to the car after lunch, Nancy asked to see the napkin. Natalie was hesitant. She finally reached into her pocket and handed her mom the napkin. Nancy unfolded it and read what she had scribbled in purple crayon: "to have my disease go away forever." Nancy's eyes began to tear up. Natalie whispered, "It's okay Mom—I'll be alright."

Natalie's wish was the catalyst in Nancy and Jeff's quest to find a cure. Those seven words provided the motivation to establish the foundation. They knew at that moment that they needed to make every effort to make Natalie's wish—and the wish of others

GoodSearch

The Cystinosis Research Foundation is listed on GoodSearch.com, a search engine that donates about a penny per search to the charities its users designate. Every time you use GoodSearch.com (instead of your current search engine, such as Google) money goes to CRF. The site is powered by Yahoo. Go to **www.goodsearch.com** and type in Cystinosis Research Foundation in the "my charity" box. Tell your friends. If 1,000 people search twice a day for a year, CRF could earn $7,300!

with cystinosis—a reality.

Cystinosis, a rare, metabolic and fatal disease, afflicts about 2,000 people worldwide. Today, Natalie's wish has also become a powerful rallying cry for friends and supporters of the Cystinosis Research Foundation (CRF) from around the world.

Natalie's wish, now known as the "wish heard round the world" resonated loud and clear at this year's Natalie's Wish event on Saturday, May 21 at the Balboa Bay Club in Newport Beach, Calif. CRF founders Jeff and Nancy Stack welcomed more than 335 guests including 32 cystinosis families and 18 renowned doctors and scientists from around the world who gathered to celebrate the foundation's most recent successes. While those accomplishments have already dramatically improved the lives of many cystinosis patients, guests also heard about developments—extraordinary life-saving developments—that hold the promise of becoming reality in the near future.

"CRF-funded research at The Scripps Research Institute has resulted in extraordinary progress moving us closer to a cure," Nancy told guests. "Researchers have been able to reverse cystinosis in mice using bone marrow stem-cell transplantation. After mice were transplanted, every single organ in the mouse saw a reversal of cystinosis."

No less impressive, some of the discoveries made by CRF-funded researchers are also being tested as treatments for Huntington's disease, Parkinson's disease and NASH, a progressive liver disease.

Of course, most of the research that is bringing so much hope to the cystinosis community and others would not be possible without the generosity of friends and supporters who contributed $1.6 million at this year's event. Their gifts bring CRF's 9-year total to almost $16.4 million, making the CRF the leading funding source for bench and clinical investigations for cystinosis. CRF has awarded research grants in eight countries and funded 78 multi-year studies and 13 research fellows worldwide. Because the foundation's administrative costs are privately underwritten all donated funds are dedicated to research.

Three leading CRF-funded researchers and a pharmaceutical company were honored on May 21:

**Corinne Antignac, MD, PhD,** of Necker Children's Hospital in Paris, whose pioneering work led to the identification of the cystinosis CTNS gene.

**Ranjan Dohil, MD,** professor of pediatrics at the University of Calif., San Diego, who with **Dr. Jerry Schneider,** developed a delayed-release form of cysteamine, which is the only cystinosis medication used to treat the disease.

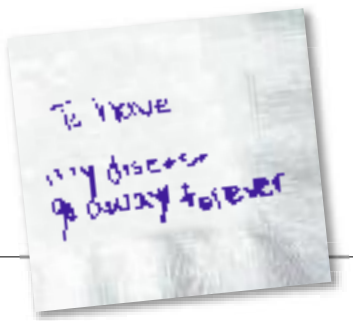
**Stephanie Cherqui, Ph.D.,** assistant professor at The Scripps Research Institute in San Diego, Calif., who established proof of concept for bone marrow stem-cell transplantation in the mouse model for cystinosis, in essence reversing cystinosis in cystinotic mice.

**Raptor Pharmaceuticals Corp.** of Novato, Calif., led by President Ted Daley, for guiding a new delayed-release medication through the grueling FDA approval process.

Finally, guests were mesmerized as they listened as a strong but almost tearful Denice Flerchinger told how her daughter Tina faces life with the joy, excitement and anticipation of a typical 7-year-old despite enduring a harrowing 24/7, 365 days a year, medical regimen.

Near the end of the evening the audience exploded with ovation when young cystinosis patients joined master of ceremonies Tony Award-winner Tim Kashani and the sensational pop band, Overtone on stage as they all sang the evening's theme song, *When You Wish Upon a Star.*

**More** For more information on cystinosis or to learn how you can help us cure this disease once and for all, go to www.natalieswish.org



**Cystinosis Research Foundation Fellowship Program**

The Cystinosis Research Foundation has established a postdoctoral research fellowship program to attract qualified, promising investigators to establish careers in cystinosis research. Fellows will be funded for 2 to 3 years to a maximum of $75,000 per year. Applications will be available in conjunction with the spring and autumn Call for Funding Proposals. Visit www.cystinosisresearch.org for details.


# Please Join Us Monday, November 14, 2011

Nancy and Jeff Stack, and Golf Chairman Vince Ciavarella, invite you to join them at the 2011 Fore a Cure Golf Tournament, on Monday, November 14. This year's tournament will be held at the exclusive Santa Ana Country Club in Santa Ana, California. Previous tournaments have sold out while raising $630,000 for cystinosis research.

**Live auction items include relaxing getaways, fine wines, dinners, and many other exciting gifts. Here are some of our donors and their generous donations:**

- Carolyne and Kim Megonigal – a Honda Center suite and 10 tickets for the November 20 game between the Ducks and Red Wings
- Kris Kristjansson – round trip airfare, accommodations and tour for four of the Jack Daniel's Distillery in Tennessee
- Jill and John Manly – a long weekend stay for 12 people at Vista Las Palmas in Palm Springs
- Nancy and Jeff Stack – an "exceptional case" of wine from their private wine cellar
- Chef Alan Greeley and Nancy and Jeff Stack – dinner for eight at the Golden Truffle restaurant with wine provided by the Stacks
- Mary and Peter Rooney – a Duffy Boat cocktail cruise followed by dinner at the Bluewater Grill
- Lynne and Augie Nieto – an Octane xR3 stationary seated exercise bike

CYSTINOSIS RESEARCH FOUNDATION

**www.cystinosisresearch.org**

**Sponsorship opportunities are still available. For information contact Zoe Solsby at 949-809-2400 or zsolsby@sares-regis.com.**

## Thank you to our generous sponsors

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# *It is better to give than to receive.*

*The old adage, always so true, is never more so than when the gift you are giving is the gift of a healthier, happier and longer life.*

*As the holiday season approaches, we invite you to consider making a donation to the Cystinosis Research Foundation on behalf of your family, your children or grandchildren, or some other family member or loved one or your company and its associates and customers.*

*The research being conducted today is some of the most promising and exciting in the history of CRF. Since every dollar donated goes directly to research, every gift, large or small, holds the possibility of being the one that pushes us over the top to ensure that all of those with cystinosis have long and happy lives free from this dreaded disease.*

*Cystinosis Research Foundation will be happy to send an acknowledgment letter to the appropriate person, should you make a donation in honor of an organization, family member or individual.*

CYSTINOSIS
RESEARCH FOUNDATION

**To make a gift to the Cystinosis Research Foundation, call 949-223-7610 or visit www.cystinosisresearch.org.**

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

# Good Bye Dear Friend: Unforgettable Memories of David Brink

The Brink brothers, Bill and Dave, were simultaneously diagnosed with cystinosis at the ages of 2 and 4. We lost Dave to cystinosis on Easter Sunday, 2011, two weeks shy of his 31st birthday.

Dave was very active in planning his funeral and wrote the following in his preparation for that day. We share this with the cystinosis community because it sets the stage for who this remarkable young man was. How wonderful for us that it came from his words. What a wonderful gift he gave us. Dave is now in a better place; whole once again, doing the things he loved.

We thank the Cystinosis Research Foundation for all of its hard work in finding better treatments and a cure for cystinosis. You give us tremendous hope.

The Brinks, Will, Joyce and Bill, Union, Kentucky

*By David Brink*

I was born in Northern Kentucky and have lived here all my life. I learned to fish at a very early age. I would never give up. When no one else could catch a fish I would still continue to cast my pole. Consequently, I would also catch the most and the largest fish. By the age of four I could out fish my parents and my aunts and uncles. One of my first memories was fishing at my Great Uncle PR and Great Aunt Kate's with cousin Laura and brother Bill. As I grew older I would continue to set goals and say to myself, "I must succeed."

**"I am glad I will not be young in a future without wilderness."**
**– ALDO LEOPOLD**

At the first house we lived in there was a creek in the backyard. Dad made us a concrete dam to play on, but we rarely used it, as we liked to get dirty and play in the creek.

We would carry empty coffee cans to catch crawdads and an occasional salamander.

Mom would sit in a lawn chair on the dam reading a book and keeping her eye on us.

Every spring the neighbor boys, Bill and I would have an annual snake round up. We would take five gallon buckets, take turns chasing garter snakes in the bucket and release the snake down by the creek.

At the age of nine, mom and dad decided it was time put the house up for sale and move. While Bill and I would wait for the school bus, we would take the for sale sign out of the yard and lay it down, in hopes that no one would know it was for sale. But once mom and dad found a new house that had an in-ground swimming pool the sign remained up.

After moving to Wedgewood, I became good friends with Mike Puckett. He would become a playmate, schoolmate and friend for life. Mike and I achieved many things together, one being grade school bowling champs named the "Bald Eagles."

While in middle school my grandparents had a lake house at Elk Lake. They told me I'd be disappointed, because the lake had little fish. Over the years I proved them wrong. I caught many trophy fish in the years to come, some of which weighed more than five pounds.

**"I enjoy getting high on mother nature."** **– TED NUGENT**

As I became a teenager, my illness worsened. I became very ill and required a kidney transplant. I had a very difficult time coming to terms with this. I needed to learn to take care of my body, and take multiple medications that tasted bad and had terrible side effects. Yet I continued to fish. I also took up hunting as a way of relaxing and enjoying myself.

The more I hunted and fished the more I grew interested in and began to gain knowledge about wildlife and the outdoors. The more I learned – I knew this was not only a sport – the more I realized that it was something I wanted to pursue as a career after high

school. I wanted to pursue a fish and wildlife career to assure that there would be the beauty of wildlife for generations to come.

I went to Hocking College in Nelsonville, Ohio. After getting over my homesickness, I soon met men and women who were as passionate about the outdoors as I. These were some of the best years of my life. When not in school learning about wildlife management I was hunting or fishing. If not outdoors or in class, I would be talking about the outdoors with my friends. I also learned what it was to be on my own. My buddies would often ask me to make "Kentucky Slop." Mom called it "college cooking" – one pot and one dish. Dad, mom and Bill would come to visit and we hiked throughout the beautiful Hocking Hills.

It was in college that I began to notice my disease was taking a turn for the worse. I could not pass the swimming test required for one of my courses. I began to have muscle loss in my hands and upper body.

After graduating from college, I went to work at Dick's Sporting Goods in the Hunting Lodge, where I worked for several years. At Dick's I was again able to share my knowledge about the outdoors and the importance of having respect for it.

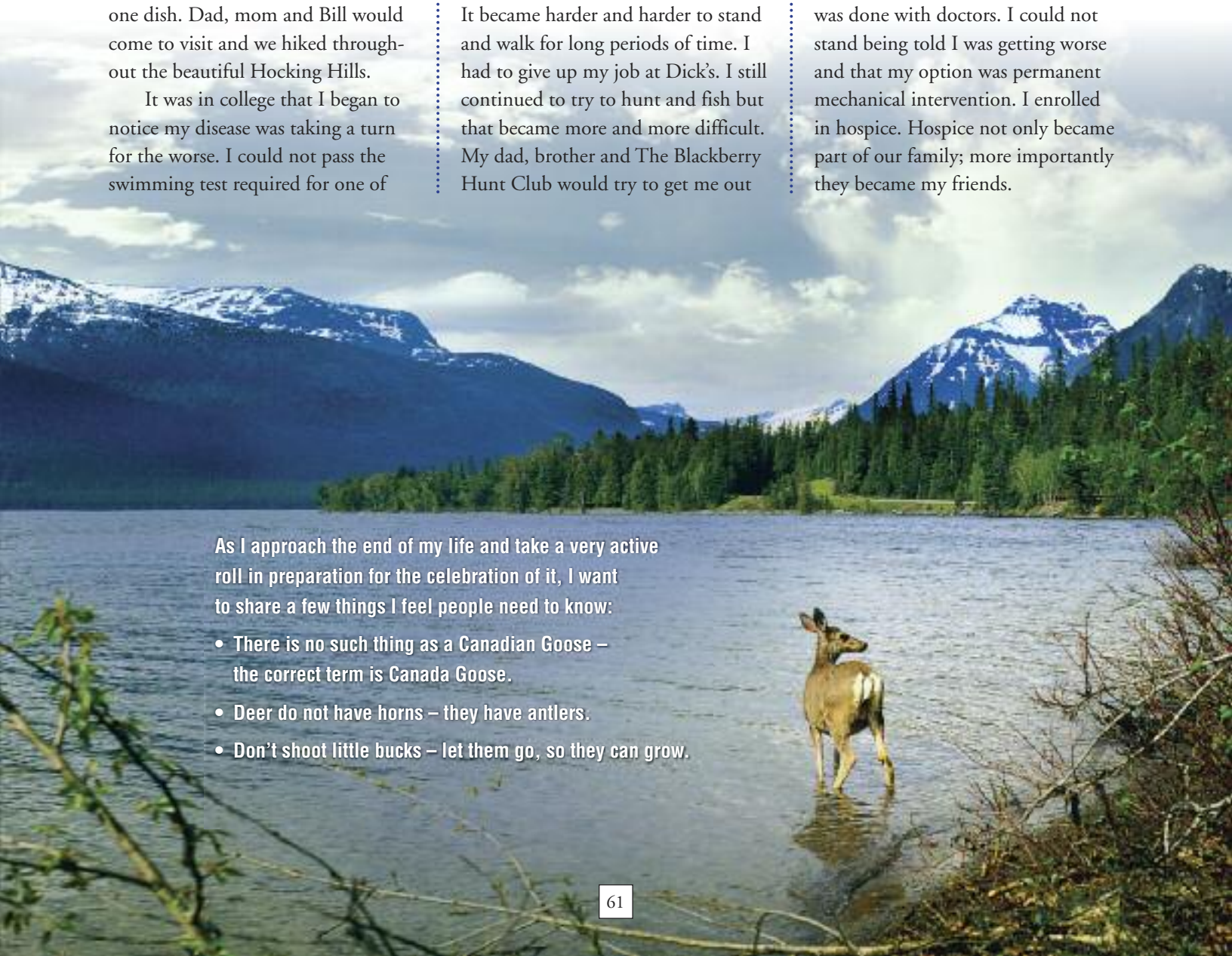
**"We abuse land because we see it as a commodity belonging to us. When we see land as a community to which we belong, we may begin to use it with love and respect."**
**– ALDO LEOPOLD**

My muscle loss worsened and I began to have problems breathing. It became harder and harder to stand and walk for long periods of time. I had to give up my job at Dick's. I still continued to try to hunt and fish but that became more and more difficult. My dad, brother and The Blackberry Hunt Club would try to get me out in the woods whenever they could and when the weather was not too severe.

Mom and Dad decided it was time to move. Dad built a new home on a lot that faced the woods. This is where I would spend most of my time the last 2 years of my life, watching deer and the many types of birds that visited our feeders.

I finally got a dog. I named her Annie, after Annie Oakley. She became a very important member of our family. Since I could no longer be outdoors I asked for an aquarium. That one aquarium quickly became three, as my African Cichlids began to breed. Our family became very close during this time.

In January of 2010 I decided I was done with doctors. I could not stand being told I was getting worse and that my option was permanent mechanical intervention. I enrolled in hospice. Hospice not only became part of our family; more importantly they became my friends.



**As I approach the end of my life and take a very active roll in preparation for the celebration of it, I want to share a few things I feel people need to know:**

- **There is no such thing as a Canadian Goose – the correct term is Canada Goose.**
- **Deer do not have horns – they have antlers.**
- **Don't shoot little bucks – let them go, so they can grow.**

# KIDNEY DONATIONS

## Did You Know?

**According to the National Kidney Foundation, there is an urgent need to increase the number of kidney donations.**

**In the cystinosis community, there are several children and young adults waiting for a kidney transplant.**

- The average wait for a kidney transplant is 3–5 years.
- Currently, there are 89,331 people waiting for a kidney transplant in the United States.
- Last year, 16,898 transplants took place in the United States.
- Of these, 10,622 kidney transplants came from deceased donors and 6,276 came from living donors.
- Every day approximately 18 people die while waiting for a life-saving kidney transplant.
- Last year, 4,636 patients died while waiting for a kidney transplant. Please give the gift of life and consider being a kidney donor!

**Become a Donor**

- Join your state's online registry for donation, if one is available.
- Declare your intentions on your driver's license.
- Sign and carry a donor card

**To learn more about kidney disease and transplantation, visit www.kidney.org.**

**If you would like to learn more about being a kidney or organ donor visit www.donatelife.net.**

# CRF Extends Global Presence

### OCTOBER 20, 2011 – SAN FRANCISCO, CA

**InnovationLab: Translating the Patient Experience into Commercial Success**

*Collaboration Amongst Industry, Academia and Advocacy: The Development of RP103*

Orphan drug development is driven by multiple parties, including academia, patient advocacy groups, government and industry. The ideal situation occurs when all these stakeholders are aligned and work together. This collaboration facilitates funding and support for research, rapid accrual of trials, appropriate community awareness of the candidates/trials, valuable input and participation by the experts, and support during the regulatory approval process.

Patrick Reichenberger, Raptor Vice President of Commercial Development and CRF President Nancy Stack shared their insights into the relationship between Raptor and CRF, which expedited development of RP103 and moved it to clinical trials.

### OCTOBER 25 – OCTOBER 29, 2011 – SÃO PAULO, BRAZIL

**IX Congress-ALANEPE Latin American Congress of Pediatric Nephrology**

CRF is a co-sponsor of the IX Congress-ALANEPE Latin American Congress of Pediatric Nephrology, which covers the latest advances in pediatric nephrology and cystinosis including panel discussions, clinical cases and poster sessions.

CRF President Nancy Stack joined leading experts in the field of cystinosis, sharing information about CRF's latest achievements and its ever-growing global research program. Nancy also discussed progress of the CCIR, which has been enthusiastically embraced by cystinosis researchers and families worldwide.

### NOVEMBER 15 – NOVEMBER 17, 2011 – BOSTON, MA

**2nd World Orphan Drug Summit –** The Summit will bring together a mix of industry leaders from pharma/biotech companies, patient advocacy groups, regulators, investors and insurance companies to share approaches, challenges and successes in orphan drug development.

CRF President Nancy Stack and Raptor Pharmaceutical Vice President Patrick Reichenberger will present a case study entitled, *The relationship between the manufacturer, biotech company and patient advocacy group prior to launch.*

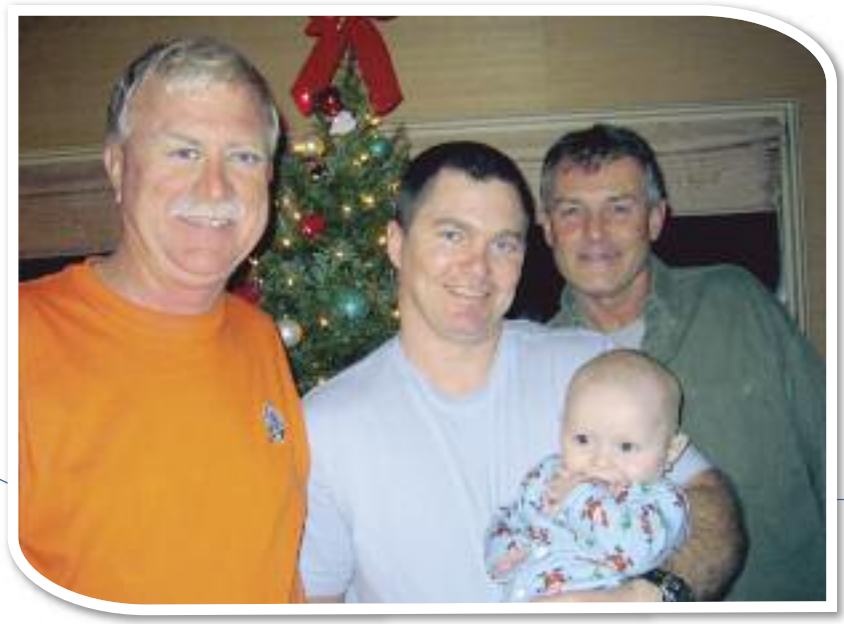
# Michael Moore

## A Special Friend of Hank Sturgis Leaves a Wonderful Legacy

Although no one at Cystinosis Research Foundation had ever heard of Michael Moore, he recently left irrefutable proof for all of us that he was an extremely kind and giving person. And he did so without sharing his plans with anyone until CRF Board Chair Nancy Stack received a wonderful, if surprising, letter in early October.

Michael was born on December 4, 1946 and he passed away on October 2, 2011 following a brief and valiant fight with metastatic brain cancer.

There is much we could share with you about Michael's life. He grew up in Clarkston, Washington before attending Washington State University where he was a member of Delta Upsilon fraternity and graduated from the school of business. Mike spent the past 25 years living in Oklahoma, but he was first and always a "Coug."



*Hank on his first Christmas Day and the first of many with his very good friend Michael Moore, his dad Brian Sturgis and his uncle Scott Landreth.*

*Mike's gift is the largest single gift CRF has received this year, and it is the first-ever estate gift in CRF history.*

Mike was well-respected throughout the country in his exclusive part of the trucking industry. He had many friends but none more special than those in Inola, Oklahoma, a small community northeast of Tulsa.

But it was his friendship with Hank Sturgis that tells us what a truly special man he was. Mike met Hank the first Christmas after Hank was born in 2006 and a year before Hank was diagnosed with cystinosis.

Tricia Sturgis fondly recalls, "Mike spent every Christmas with our family since 1987. Mike loved Hank and would always ask a lot of questions about cystinosis in general, CRF and our foundation in honor of Hank. Mike was such a generous man, happy and easy-going.

"We loved Mike too; we enjoyed spending time with him catching up on his trucking adventures and watching football. Mike had a wonderful smile and it sure showed when Hank was around.

"We will forever keep Mike in our hearts. Our family is humbled, grateful and thankful for his gift to cystinosis."

Mike wanted to help Hank even in his passing. He quietly and unexpectedly left $53,000 in his will to CRF, through the Sturgis's foundation, 24-Hours for Hank.

Mike's gift is the largest single gift CRF has received this year, and it is the first-ever estate gift in CRF history. Because of Mike's love for Hank and his concern for all those with cystinosis, Mike will forever be remembered at the Cystinosis Research Foundation.

If you would like to include Cystinosis Research Foundation in your estate planning, please contact Nancy Stack at 949-223-7610 or nstack@cystinosisresearch.org.

# Studies Published by CRF-Funded Researchers

**Quantitative in Vivo and Ex Vivo Confocal Microscopy Analysis of Corneal Cystine Crystals in the *Ctns-/-* Knockout Mouse** – *Molecular Vision Journal,* August 2011 by Jennifer Simpson, MD

**Cysteamine Toxicity in Patients with Cystinosis** – *Journal of Pediatrics,* July 2011 by Elena Levtchenko, MD, PhD

**The 57 KB Deletion in Cystinosis Patients Extends into *TRPV1 Causing* Dysregulation of Transcription in Peripheral Blood Mononuclear Cells** – *Journal of Medical Genetics,* May 2011 by Katy Freed, PhD

**Kidney Preservation by Bone Marrow Cell Transplantation in Hereditary Nephropathy** – *Kidney International Journal,* May 2011 by Brian Yeagy, PhD and Stephanie Cherqui, PhD

**Kidney Repair and Stem Cells: A Complex and Controversial Process** – *Pediatric Nephrology,* January 2011 by Brian Yeagy, PhD and Stephanie Cherqui, PhD

**Kidney Preservation by Bone Marrow Cell Transplantation in Hereditary Nephropathy** – *Pediatric Nephrology,* November 2010 by Brian Yeagy, PhD and Stephanie Cherqui, PhD

**Long-Term Treatment of Cystinosis in Children with Twice-Daily Cysteamine** – *The Journal of Pediatrics,* May 2010 by Ranjan Dohil, MD

**Novel Conditionally Immortalized Human Proximal Tubule Cell Line Expressing Functional Influx and Efflux Transporters** – *Cell Tissue Research,* 2009 by Elena Levtchenko, MD, PhD

**The Redox Status of Cystinotic Fibroblasts** – *Journal of Molecular Genetics and Metabolism,* January 2010 by Jess Thoene, MD

**Results from Phase 2a Trial of DR Cysteamine for Treatment of Cystinosis** – *Journal of Pediatrics,* January 2010 by Ranjan Dohil, MD

**Renal Phenotype of the Cystinosis Mouse Model is Dependent Upon Genetic Background** – *Nephrology Dialysis Transplantation Journal,* October 21, 2009 by Corinne Antignac, MD, PhD

**Successful Treatment of the Murine Model of Cystinosis Using Bone Marrow Cell Transplantation** – *Blood Journal,* September 17, 2009 by Stephanie Cherqui, PhD

**Heterozygous Mutation of Opa 1 in *Drosophila* Shortens Lifespan Mediated through Increased Reactive Oxygen Species Production** – Publised Plos One, February 2009 by Sha Tang and Taosheng Huang

**An Indian Boy with Nephropathic Cystinosis** – *Genetic Testing and Molecular Biomarkers,* Volume 13, Number 4, 2009 by Sha Tang and Taosheng Huang

**CRF First International Cystinosis Research Symposium** – Abstracts – *Pediatric Nephrology Journal,* September 2008

**Gene Transfer May Be Preventive But Not Curative for a Lysosomal Transport Disorder** – *Official Journal of the American Society of Gene Therapy,* July 2008 by Vasiliki Kalatzis, PhD

**Identification and Subcellular Localization of a New Cytinosin Isoform** – *American Journal of Renal Physiological,* March 2008 by Anna Taranta, PhD; Francesco Emma, MD; Elena Levtchenko, MD, PhD

**Pharmacokinetics of Cysteamine Bitartrate Following Gastrointestinal Infusion** – *British Journal of Clinical Pharmacology,* August 2006 by Meredith C. Fidler, Bruce A. Barshop, Jon A. Gangoiti, Reena Deutsch, Michael Martin, Jerry A. Schneider and Ranjan Dohil

**Understanding Intestinal Cysteamine Bitartrate Absorption** – *Journal of Pediatrics,* June 2006, Ranjan Dohil, MD

**A Deeper Look into Cysteamine Absorption for the Treatment of Cystinosis** – Editorial – *Journal of Pediatrics,* June 2006

**Treatment of YAC128 Mice and Their Wild-type Littermates with Cystaimine Does Not Lead to Its Accumulation in Plasma or Brain: Implications for the Treatment of Huntington Disease** – *International Society for Neurochemistry,* 2005 by Tom Jeitner, PhD

**Potential Role of Apoptosis in Development of the Cystinotic Phenotype** – *Pediatric Nephrology Journal,* December 2004, Jess Thoene, MD

CYSTINOSIS RESEARCH FOUNDATION

# Dr. Corinne Antignac Continues Her Extraordinary Service to the Cystinosis Community

**Dr. Corinne Antignac's** pioneering work, in collaboration with others, led to the identification of the CTNS "cystinosis" gene in 1998.

In addition to extensive research and publishing schedules, Dr. Antignac is the current chair of the CRF Scientific Review Board, a member of the CRF Cystinosis Gene Therapy Consortium and co-chair of the CRF International Research Symposium. She is also a critical "go-to" source for other cystinosis researchers, as well as CRF Board Chair Nancy Stack.

*Dr. Corinne Antignac receiving the Legion d'Honneur in 2008. The award is the highest decoration in France.*

Dr. Antignac has been a longtime friend of CRF and the cystinosis community. She presented her work at the 2011 *Day of Hope.* She is also a longtime mentor to, and colleague and friend of Dr. Stephanie Cherqui.

Dr. Antignac is a member of several national and internal societies including the Société de Néphrologie Pédiatrique, Société de Néphrologie, European Society for Paediatric Nephrology, International Society of Nephrology, American Society of Nephrology, and numerous scientific boards.

She has received numerous awards, including the Legion d'Honneur in 2008, for her remarkable body of scientific work. The Legion d'Honneur, the highest decoration in France, was established by Napoleon Bonaparte in 1802. Dr. Antignac was also recognized at our 2011 Natalie's Wish event for her leadership and dedication to the cystinosis community.

## 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.

## 2012 Call for Funding Proposals

The ultimate goal of the Cystinosis Research Foundation is to find a cure for cystinosis. Global calls for grant applications are made twice-a-year, near or around March 1 and September 1. Deadline for applications will be in April and October respectively. Research and fellowship awards will be given for up to 3 years.

Currently, CRF has more than $1 million available for grants. The number and value of awards will depend on the number of outstanding proposals and the funds available at the time. **Visit www.cystinosisresearch.org for details.**

## 2011 CRF Spring Research Grants Awarded

### Total: $1,244,355

**Bruce Barshop, MD, PhD**
University of California, San Diego
"Tandem Mass Spectrometer Support"
$121,130 – 1-year grant

**Sergio Catz, PhD, Mentor**
**Gennaro Napolitano, PhD, Research Fellow**
The Scripps Research Institute, La Jolla, California
"Small Molecule Regulators of Vesicular Trafficking to Enhance Lysosomal Exocytosis in Cystinosis."
$129,050 – 2-year grant

**Stephanie Cherqui, PhD, Principal Investigator**
The Scripps Research Institute, La Jolla, California
"Mechanism of Bone Marrow Stem Cell-Mediated Therapy in the Mouse Model of Cystinosis."
$401,574 – 2-year grant

**Pierre Courtoy, MD, Principal Investigator**
**Heloise Chevronnay, PhD, Co-Principal Investigator**
De Duve Institute, Brussels, Belgium
"Cellular and Tissular Mechanisms for Stem Cell Therapy of Epithelial Cells in a Mouse Model of Cystinosis Coping with Tissue Heterogeneity."
$126,618 – 1-year grant

**Ranjan Dohil, MD**
University of California, San Diego
"Two Year Funding Proposal for Betty Cabrera, BS, MPH."
$194,250 – 2-year grant

**Ranjan Dohil, MD**
University of California, San Diego
"A Study to Evaluate Enteric-Coated Cysteamine Therapy in Patients with Cystinosis"
$52,767 – 2-year grant

**Daryl Okamura, MD, Principal Investigator**
**Allison Eddy, MD, Co-Principal Investigator**
Seattle Children's Research Institute, Washington
"Elucidating the Role of Cystinosin-Deficient Macrophages in Nephropathic Cystinosis."
$218,966 – 2-year grant

# Cystinosis Research Foundation

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

## BOARD OF TRUSTEES

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**Jody Strauss**
Cystinosis Awareness and Research Effort

**Tricia Sturgis**
24 Hours for Hank

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**Jennifer Simpson, MD**
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Irvine, California

**Doris Trauner, MD**
University of California, San Diego
San Diego, California

## MISSION

The mission of the Cystinosis Research Foundation is to support bench and clinical research that is focused on developing improved treatments and a cure for cystinosis.

## EDUCATION

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

## 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.

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

**Stephanie Cherqui, PhD**
The Scripps Research Institute
La Jolla, California

**Allison A. Eddy, MD**
Seattle Children's Hospital
Seattle, Washington

**Francesco Emma, MD**
Bambino Gesù Children's Hospital
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**Julie R. Ingelfinger, MD**
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University of Nebraska Medical Center
Omaha, Nebraska

CYSTINOSIS
RESEARCH FOUNDATION

**Zoe R. Solsby,** Vice President

# John Manly, Dr. Stephanie Cherqui and Jody Strauss Join CRF Board

**John Manly** is a leading civil authority on the clergy abuse crisis in the United States and abroad. He has been prominently featured in print and on broadcast news in the U.S. and abroad for exposing perpetrators and the men and women who cover up for them. He has recovered hundreds of millions of dollars for the victims.

In 2005, John appeared in Amy Berg's Academy Award-nominated documentary, *Deliver Us From Evil,* which featured his role in the civil prosecution of predator priest Oliver O'Grady and the church officials who allowed O'Grady to continue abusing children.

Born in San Mateo and raised in Southern California, John obtained a bachelor's degree from the University of Southern California and his J.D. from Pepperdine University Law School.

John is also a well known expert on the legal issues facing multifamily and commercial properties. He has addressed national conventions of the largest property owners and managers in the country. He has written numerous articles about real estate and construction law which have been published in the industry's top trade publications.

**Dr. Stephanie Cherqui** is an assistant professor at The Scripps Research Institute in San Diego, California. The main focus of her laboratory is the use of bone marrow stem cells for kidney repair and for the treatment of cystinosis.

She obtained her PhD in 2002 from the University Rene Descartes, Paris, France while working in the laboratory of Dr. Corinne Antignac. While there she participated in the identification of the CTNS gene, the cystinosis gene.

Dr. Cherqui joined Dr. Daniel Salomon's group at The Scripps Research Institute in 2002 for post-doctoral training on stem cells and gene therapy using lentiviral vectors applied on vascular diseases. In 2006, she started the project "Stem cells and gene therapy for cystinosis." Using the mouse model for cystinosis, she showed that transplantation of bone marrow stem cells or hematopoietic stem cells expressing a functional Ctns gene results in the abundant tissue engraftment of transplanted cells and decrease of cystine content.

Dr. Cherqui is a member of the CRF Scientific Review Board, chair of the CRF Cystinosis Gene Therapy Consortium and co-chair of the CRF International Research Symposium. She is also a member of the American Society of Gene and Cell Therapy.

**Jody Strauss** deals with the realities of cystinosis on a daily basis. Her 3 ½ year old daughter, Gabbie was diagnosed with cystinosis in 2009. Shortly after Gabbie's diagnosis Jody and her husband Trevor established the Cystinosis Awareness and Research Effort (CARE), the first non-profit of its kind in Canada. The first year of fundraising, with more than 10 events, included grass roots bake sales, association golf tournaments and large scale community events. Since its inception, CARE has raised over $200,000 for cystinosis research.

Prior to Gabbie's birth Jody spent 5 years working in public health, spear-heading no-smoking legislation and implementing heart health and colon screening programs in Ontario. Jody is involved in her faith community and is a regular volunteer in the Kingdom Kidz program at Waterloo Mennonite Brethren Church.

Jody is committed to helping children and families affected by cystinosis. She hopes to inspire and build momentum amongst Canadian cystinosis families to raise awareness and research funds in their respective communities.

Jody has a Honours Bachelor of Applied Science in Human Nutrition from the University of Guelph, in Guelph, Ontario. Jody is happily married to husband Trevor and lives in Waterloo, Ontario, Canada with her daughters Gabbie and Chloe and their family dog, Sadie.

# CURE CYSTINOSIS INTERNATIONAL REGISTRY

**The Cure Cystinosis International Registry (CCIR) is a collaborative effort by the leaders in the cystinosis community to establish a comprehensive, global patient registry for cystinosis. The purpose is to connect those with cystinosis to the research community in an effort to find a cure for cystinosis.**

## CCIR BOARD OF ADVISORS

The CCIR Advisory Board is dedicated to promoting and facilitating current research and medical information to the global cystinosis community in an effort to inform the community of current treatments, clinical trials and studies and patient care.

**Bruce A. Barshop, MD, PhD**
University of California, San Diego

**Kyle Brown,** CEO, Innolyst, Inc.

**Betty L. Cabrera, MPH**
University of California, San Diego

**Paul Goodyer, MD**
Montreal Children's Hospital, Canada

**Whitney Glaize**
Cystinosis Research Foundation

**Jean Hobbs-Hotz,** Cystinosis Foundation

**Valerie Hotz,** Cystinosis Foundation

**Tom Melang,** Cystinosis Patient Advocate

**Patrick Niaudet, MD**
Hôpital Necker-Enfants Malades,
Université Paris-Descartes, Paris, France

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

**Jerry Schneider, MD**
Dean for Academic Affairs, Emeritus
University of California, San Diego

**Nancy Stack,** Cystinosis Research Foundation

**Tricia Sturgis,** 24 Hours for Hank

**Doris A. Trauner, MD**
University of California, San Diego

## CCIR MEDICAL AND SCIENTIFIC COUNCIL

The CCIR Medical and Scientific Council was instrumental in the development, design and content of the medical questionnaire for the registry. The Council provides ongoing guidance relating to the scientific and clinical aspects of the registry.

In addition, Dr. Barshop, Dr. Goodyer, Dr. Schneider and Dr. Trauner are members of the *Ask An Expert* sub-committee that addresses questions from cystinosis patients.

**Bruce A. Barshop, MD, PhD**
University of California, San Diego

**Stephanie Cherqui, PhD**
The Scripps Research Institute, La Jolla

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

**Paul Goodyer, MD**
Montreal Children's Hospital, Canada

**Daniel R. Salomon, MD**
The Scripps Research Institute, La Jolla

**Jerry Schneider, MD**
Dean for Academic Affairs, Emeritus
University of California, San Diego

**Doris A. Trauner, MD**
University of California, San Diego

**www.cystinosisregistry.org**

# Third International Symposium to Drive Collaboration on the Push Toward a Cure

THIRD INTERNATIONAL
Cystinosis Research Symposium

**At the Second CRF International Cystinosis Research Symposium held in April 2010, more than 65 leading researchers and scientists from eight nations gathered to share information about their research and exchange ideas about promising new study directions, all with the goal of finding better treatments and ultimately a cure for cystinosis.**

Based on the enormous success of the 2008 and 2010 conferences, renowned cystinosis researchers from around the world will again gather to present their research in an effort to accelerate treatments and a cure. The two-day event will take place on Thursday, March 29 and Friday, March 30, 2012 at the Arnold and Mabel Beckman Center of the National Academies of Sciences and Engineering in Irvine, California.

The conference is again organized and underwritten by the Cystinosis Research Foundation.

Dr. **Jerry Schneider**, with his long-standing commitment to cystinosis research, is the symposium's honorary chair. Dr. Schneider chaired the previous two conferences. That role in 2012 is being filled by four prominent scientists: Dr. **Corinne Antignac**, Hôpital Necker-Enfants Malades, Paris, France; Dr. **Stephanie Cherqui**, The Scripps Research Institute, La Jolla, California; Dr. **Julie Ingelfinger**, Harvard Medical School, Boston, Massachusetts; and Dr. **Elena Levtchenko**, University Hospital Leuven, Leuven, Belgium.

In addition to extensive research and publishing schedules, the conference co-chairs all serve on the CRF Scientific Review Board (SRB), which Dr. Antignac chairs. The SRB has the challenging job of evaluating every research proposal submitted to CRF during the Foundation's twice-a-year calls. In both 2010 and 2011, the SRB reviewed approximately 30 proposals from scientists around the world.

**Third CRF International Cystinosis Research Symposium**

**Thursday, March 29 and Friday, March 30, 2012**

**THE SYMPOSIUM WILL:**

- ★ Highlight research being carried out by cystinosis investigators
- ★ Facilitate the exchange of knowledge
- ★ Enhance participants awareness of global research progress
- ★ Foster research collaborations

**BECKMAN CENTER OF THE NATIONAL ACADEMIES OF ENGINEERING AND SCIENCE**
Irvine, California 92617

For more information contact **Nancy Stack** at **949-223-7610** or **nstack@cystinosisresearch.org**.

Once again, the openness, collaboration and camaraderie, generally rare at conferences attended by high-profile researchers, are expected to drive participation at next year's event. Many attendees claim that the synergy in the cystinosis research community which they believe plays a key role in the success of their individual efforts, is a direct outgrowth of the symposium.

"Dissension rules other scientific worlds, but collaboration describes the ethic of the cystinosis community," said **William A. Gahl**, MD, PhD, clinical director of the National Human Genome Research Institute.

"All the researchers who attend are passionate about their research and they are committed to our children and dedicated to understanding more about cystinosis," said **Nancy Stack**, co-founder of the CRF with her husband, Jeff. Nancy also chairs the CRF Board of Trustees. "Once again we are certain that cystinosis scientists will share their data and discoveries, discuss new theories and form collaborations. Accelerating research and outcomes is one of the primary reasons we sponsor the symposium."

Gene therapy is one of the most promising areas of research and a focus of CRF funding. **Donald Kohn**, MD, Professor, Microbiology, Immunology and Molecular Genetics and Pediatrics at UCLA and a member of the CRF Gene Therapy Consortium will speak at the plenary session. "We are dedicated to bringing the first stem cell and gene therapy treatment for cystinosis to reality – we are making enormous advances," said Stack.

**If you would like more information about the symposium contact Nancy Stack, Foundation Chair, at 949-223-7610 or nstack@cystinosisresearch.org.**

# TOGETHER WE ARE MAKING A DIFFERENCE. *Save the Dates*

**Sunday, November 6, 2011 ★ 11 am – 2pm**
Cut-A-Thon to benefit CRF in honor of Glenn Jones
***Jon Francis & Co – (561) 443-2055***
Boca Raton, Florida

**Monday, November 14, 2011 ★ 10:30 am**
Cystinosis Research Foundation
***Natalie's Wish Fore a Cure Golf Tournament***
Santa Ana Country Club, Santa Ana, California

**Friday, January 27, 2012**
Cystinosis Awareness and Research Effort – Gabbie Strauss
***Gourmet Soup & Cracker Event***
***Dana Shortt Gourmet***
Waterloo, Ontario, Canada

**Saturday, February 4, 2012 ★ 7 pm**
Hope for Holt Foundation – Holt Grier
***Fifth Annual Hearts for Holt Gala***
Charlotte Country Club,
Charlotte, North Carolina

**Thursday, March 22, 2012**
Jenna and Patrick Foundation of Hope –
Jenna and Patrick Partington
***Broker of the Year Awards, Association of Commercial Real Estate***
Sacramento, California

**Saturday, March 24 – Sunday, March 25, 2012**
24 Hours of Sun Valley – Henry Sturgis
***Ski-A-Thon***
Sun Valley, Idaho

**Thursday, March 29 – Friday, March 30, 2012**
Cystinosis Research Foundation
***Third CRF International Cystinosis Research Symposium***
Beckman Center of the National Academies
of Engineering and Science
Irvine, California

**Friday, March 30, 2012**
24 Hours for Hank – Henry Sturgis
***The Fourth Annual 24 Hours of Schweitzer***
Schweitzer Mountain Resort, Sandpoint, Idaho

**Thursday, April 19 – April 21, 2012**
Cystinosis Research Foundation
***Day of Hope Family Conference***
Balboa Bay Club, Newport Beach, California

**Saturday, May 19, 2012 ★ 6 pm**
Tina's Hope for a Cure – Tina Flerchinger
***Fourth Annual Wine, Stein & Dine***
***Rogers Toyota Scion Showroom***
Lewiston, Idaho

**August, 2012**
Cystinosis Research Foundation
***Crash and Crush for a Cure sponsored by Ben's Asphalt***
Orange County Fair – Demolition Derby
Costa Mesa, California

**Saturday, September 8 – Sunday, September 9, 2012**
24 Hour Bike Ride – Henry Sturgis
***Cycling for Cystinosis***
Sandpoint, Idaho

**Connect with us at www.cystinosisresearch.org or at www.facebook.com/CystinosisResearchFoundation**

## SAVE THE DATE FOR THE 2012

# *Natalie's Wish Celebration*

Saturday, April 21, 2012

Balboa Bay Club, Newport Beach, California

THE YEAR'S MOST MAGICAL CELEBRATION.