

# Cystinosis

MAGAZINE

SPRING 2011

FOR FRIENDS AND SUPPORTERS OF THE CYSTINOSIS RESEARCH FOUNDATION



Natalie Goes to Washington  
Much Has Changed in 43 Years  
The Wish Heard Round the World  
International Registry Goes Live

**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.

Although medication is available to control some of the symptoms of this insidious disease, 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 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 around the world.



**For more information about the Cystinosis Research Foundation, or to support its mission visit [www.cystinosisresearch.org](http://www.cystinosisresearch.org) or call 949-223-7610**

18802 Bardeen Avenue, Irvine, CA 92612-1521

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

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 78 studies and fellowships in eight countries.

CRF has raised nearly \$15 million, which has funded or been 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 sufferers and their families.

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# Day of Hope

Cystinosis Research Foundation Family Conference

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

*Learn ✨ Laugh ✨ Share ✨ Celebrate*

**Join us for three exciting days at the Cystinosis Research Foundation Day of Hope Family Conference and Natalie's Wish Event on May 19 – May 21 on the beach at the Balboa Bay Club in Newport Beach, California.**

- Top cystinosis researchers from around the world will share their research progress on gene and stem cell therapies, novel eye research and neurological issues related to cystinosis.
- Raptor Pharmaceuticals will provide an update on the RP103 delayed-release medication plus Sigma-Tau will discuss the eye drop progress with the FDA.
- And cystinosis families from around the world will meet, socialize and share their personal stories of life with cystinosis and 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.**

*Fun in the Sun  
for the Entire Family*



**CYSTINOSIS**  
RESEARCH FOUNDATION  
[www.cystinosisresearch.org](http://www.cystinosisresearch.org)

For more information about the conference or the hotel, visit [www.cystinosisresearch.org](http://www.cystinosisresearch.org) or contact: Nancy Stack at 949-223-7610 or [nstack@cystinosisresearch.org](mailto:nstack@cystinosisresearch.org)

# Dear Friends and Family

**Spring is here and we feel a sense of renewed purpose and energy at the Cystinosis Research Foundation. Since its inception in 2003, we have soared to great heights and achieved significant research milestones.**

The landscape of cystinosis research has changed dramatically since the CRF was established. Today, the field is rich with novel research discoveries, new scientists committed to cystinosis research and the reality of new therapeutic treatments on the horizon. There is no doubt that the CRF has been the leading factor in bringing about this change in the field of cystinosis research.

The CRF began with the personal story of our then eleven-year-old daughter, Natalie, who made her birthday wish on a paper napkin while at lunch. Her wish, **to have my disease go away forever** was an emotional plea that could not be ignored. What struck and saddened us most about her wish was how cognizant she was about her own health and destiny. Like all children with cystinosis, our children lose their childhood innocence quickly as the realities of dealing and coping with cystinosis becomes an undeniable and brutal way of life. **Simply put, life is not easy for our children.**

In order to cope with the daily challenges of cystinosis, many parents of children with cystinosis shield their emotions and exude a sense of normalcy. However, **underneath the often calm demeanor, there is underlying fear and sense of desperation about what the future holds for those who suffer from this devastating disease.**

As you read the family stories in this magazine, I am certain that you will feel the powerful sense of optimism, courage and faith every story conveys. As a community we are sustained by hope as a result of ongoing cystinosis research and its promise to yield new treatments for our children. Void of hope, we might crumble.

Before Elizabeth Edwards passed away, she shared her feelings about her life – the words are poignant and universal. She stated she had “three saving graces – my family, my friends and a faith in the power of resilience and hope.”

The other day, as I kissed my sweet daughter good-bye at the airport as she headed back to Georgetown University, I was reminded of the fragility of life, of how much I love and miss her every day, of how scared I am for her because her health is deteriorating and yet, how proud I am of her for her personal determination and resilience.

**I was comforted, however, by the knowledge that we are on a direct path to a cure and we are getting closer every day because of the breadth and quality of cystinosis research the CRF supports. Your commitment has enabled us to fund research that will save Natalie and the other children and adults with cystinosis.**

What began as one family’s journey is now the journey of an international community. The CRF funds research around the world and we are connected to families from all over the world by our common quest – to find a cure for cystinosis.

Blessed by your love and commitment to the Cystinosis Research Foundation, together we will determinedly and resolutely make Natalie’s wish – *to have my disease go away forever* – a reality for every person with cystinosis. The journey continues and we have work to do but with your unwavering support, we will realize our collective wish and find a cure for cystinosis.

**You, my family, my friends have sustained us and are my saving graces.**

**Enjoy our inaugural issue of *Cystinosis Magazine* and know that you have been an integral part of all we have accomplished and will be part of the milestones we have yet to reach.**

With love and a grateful heart,

*Nancy and Jeff*



*Dear Friends:*

Last year I transferred from Loyola Marymount University in Los Angeles to Georgetown University in Washington DC. I love Georgetown; it is the perfect fit for me! It is an academically challenging school so I am studying harder than ever. I love the people, the community, the location and the Georgetown school spirit! Although the weather is much different from California, I enjoy the change of seasons. I like the snow and rain, so it does not bother me when the weather is challenging. In fact, I recently experienced my first “snow day” and all my classes were cancelled! I am happy I transferred – it was the right decision. Surprisingly, living across the country and away home has not been too difficult.

Although I am very happy at Georgetown, it is always nice to go home. I was there for Thanksgiving and Christmas and I enjoyed seeing and spending time with my family, friends, my cat Max and puppy, Dillon.

I am currently a second semester sophomore at Georgetown. My major is psychology and my minor will soon be English. So far, my favorite subjects are psychology, sociology and French.

I live in the dorms and have my own room, which makes it much easier for me to take my medicine and to maintain the sleeping pattern I am used to.

Since I have been on the experimental delayed-release medication, it is easy to take my medicine every day and on schedule. I take eight different medications but I only need to take them every 12

hours. And since I usually do not start classes until after 11 a.m., I don't miss any doses. Since it is hard for me to get blood draws while at school, my doctor reduced the number of draws so that I get my lab work done when I go home. I also schedule all of my doctor appointments when I am home for a visit.

I'm looking forward to 2011, and hoping the delayed-release medication will be available this year for everyone who has cystinosis. For me, the slow-release medication is a “miracle drug.” It has changed my life. Taking it less frequently has made my life much easier and means I have fewer side effects.

**Thank you all for your love and support. We would not be where we are today without you and your commitment. I greatly appreciate your generosity and help in finding a cure for cystinosis.**



Love, Natalie

# Update

## Stephanie Cherqui, PhD

Assistant Professor,  
Department of Molecular  
and Experimental Medicine,  
The Scripps Research Institute  
La Jolla, California



**Dr. Stephanie Cherqui was part of Dr. Corinne Antignac’s lab in Paris, France and participated in the discovery of the cystinosis gene in 1998. In 2002, she joined Dr. Daniel Salomon’s group at The Scripps Research Institute. In 2006, she commenced the CRF-funded study “Stem Cell and Gene Therapy for Cystinosis,” which has led to new discoveries and possible therapeutic treatments for cystinosis.**

In 2010, Dr. Stephanie Cherqui received a grant from the National Institutes of Health (NIH), the nation’s medical research agency, for her work on cystinosis. The grant is an NIH Research Project grant (RO1) for 5 years supported by the National Institute of Diabetes and Digestive and Kidney diseases.

This grant, in addition to those from the Cystinosis Research Foundation, will support the preclinical studies for the autologous transplantation of bone marrow stem cell genetically modified to express a functional *CTNS* gene. Autologous transplantation is the use of the patient’s own stem cells that have to be genetically modified *ex vivo* to introduce a functional *CTNS* gene. The gene will be introduced using a lentiviral vector that will stably integrate into the patient’s genome. The optimization of such a treatment, as well as the efficiency and safety, is currently tested in the mouse model of cystinosis – the *Ctns*<sup>-/-</sup> mice.

The premise for this work was established by Dr. Cherqui and her team who showed that transplantation of healthy bone marrow stem cells led to a significant decrease of cystine in all the tissues tested, as well as prevention or treatment of tissue injury in the *Ctns*<sup>-/-</sup> mice.

Dr. Cherqui and her team also published an article in the *Kidney International* journal on the long-term effect of bone marrow stem cell transplantation specifically on kidney injury, in the mouse model of cystinosis. Dr. Cherqui showed that bone marrow stem cells expressing a functional *Ctns* gene lead to the significant decrease of cystine levels and cystine crystals in all the tissues tested for the entire life span of the *Ctns*<sup>-/-</sup> mice. Thus, these new results suggest that this treatment could be stable for the life of the patient.



Dr. Cherqui has recently published articles in three prestigious scientific journals as a result of CRF-funded grants. The first was in the September 2009 edition of the *Blood* journal (pictured lower left). In January, 2011 her article, “Kidney preservation by bone marrow cell transplantation in hereditary nephropathy” was published in *Kidney International*. Her most recent article, published in February 2011 in *Pediatric Nephrology*, was a review of kidney repair and stem cells.



However, the *Kidney International* article describes how bone marrow stem cell transplantation could lead to the long-term preservation of the kidney in the *Ctns*<sup>-/-</sup> mice, but this necessitates a high level of stem cells expressing a functional *Ctns* gene in the bone marrow. Finally, they showed that the preservation of the kidney was not dependant of the age of the mice at the time of transplant. This data suggests that bone marrow transplantation could save a damaged kidney if the injury is not too advanced.

These studies will have to be carefully considered when designing future clinical trials using adult bone marrow stem cells for cystinosis. Translation to humans will be done with the expertise of well-established researchers and physicians within the CRF Cystinosis Gene Therapy Consortium (see next page) and should lead to the first clinical trial of stem cell and gene therapy for cystinosis.

# CRF Cystinosis Gene Therapy Consortium

A group of leading researchers in stem cell and gene therapies, molecular biology and clinical pathophysiology have formed the CRF Cystinosis Gene Therapy Consortium to find successful therapies for cystinosis.

Pre-clinical investigation by members of the consortium at The Scripps Research Institute, La Jolla,

for cystinosis to reality. If all goes well, we hope there will be a clinical trial for a cure within the next two to four years,” said consortium member Nancy Stack, a co-founder and trustee of the CRF. She added that the initial published findings also have “far-reaching implications for application to other diseases with systemic defects similar to cystinosis.”

In other human disorders, a person’s own stem cells have already been used safely, and the stem cells could target several tissues.

This new consortium was created based on the promising pre-clinical studies performed in the mouse model for cystinosis by Dr. Cherqui’s laboratory team. Transplantation of healthy bone marrow stem cells led to a significant decrease of cystine in all the tissues tested as well as prevention or correction of tissue injury.

According to Dr. Cherqui, “The critical next step is to develop the strategies necessary to successfully deliver the CTNS gene to patients’ bone marrow stem cells using gene therapy.

Of course, the translation of these studies to humans will require extensive safety studies, as well as the expertise of members of the CRF Cystinosis Gene Therapy Consortium.”

*The goal of the CRF Cystinosis Gene Therapy Consortium is to advance progress on the most promising current findings, including moving novel therapeutic modalities into human patients as quickly as possible.*

California, has resulted in the significant decrease of cystine in all tissues, as well as the prevention or treatment of tissue injury in laboratory mice, a result recently published in the journal *Blood*. The goal of the CRF Cystinosis Gene Therapy Consortium is to advance progress on the most promising current findings, including moving novel therapeutic modalities into human patients as quickly as possible. Work is now under way at The Scripps Research Institute to develop the necessary preclinical animal model data and translate these results into an FDA-approved clinical trial.

The studies are being funded privately by the Cystinosis Research Foundation, (CRF). “We are dedicated to bringing the first stem cell and gene therapy clinical trial

Principal Investigator Dr. Stephanie Cherqui explained, “Gene therapy adds a functional copy of the faulty gene and delivers it to the appropriate cells of the body. In the case of cystinosis, most of the tissues are damaged because of the lack of the CTNS gene.

## 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

# Q&A with Ted Daley

President, Raptor Therapeutics, Inc.



## 1 What is the current status of the Phase 3 RP 103 clinical trial? Specifically, when will Phase 3 end?

We are still enrolling patients in the phase 3 clinical trial. We expect to complete enrollment in March, and to have all patients complete the study by the end of May, 2011.

## 2 With data available from Phase 3, can you tell us what the benefits are of taking RP 103 vs. Cystagon®?

Although we don't have results reported from our phase 3 study yet, the study was designed to demonstrate that patients are able to control their disease, as measured by WBC cystine levels, on a twice-daily dosing regimen as compared to every six hours with the current standard of care. We also hope to demonstrate improved tolerability compared to the currently available immediate release formulation. These findings would be consistent with the results of Dr. Dohil's and Dr. Schneider's studies, and with our previously reported phase 2b study of DR Cysteamine in cystinosis patients. It is hoped that both of these outcomes would improve compliance and quality of life.

## 3 Why must RP 103 be taken with coke and orange juice but not with milk? Will this be the suggested protocol when the drug is approved?

RP103 was formulated to pass intact through the typically acidic environment of the stomach, and to dissolve in the more neutral environment of the small intestine. We have recommended that patients in our clinical study take RP103 with juice or a soft drink (which are acidic), and to avoid taking with milk or dairy products (which are not acidic), to minimize the possibility of dietary interference with the enteric coating. It is possible that the eventual label could include some dietary guidance such as a recommendation to avoid taking the drug with milk, but we won't know until the product is approved.

## 4 Once Phase 3 is complete, we understand that there will be a Phase 4 study. Would you explain what the Phase 4 study is and who will be eligible? Will post-transplant patients be eligible?

"Phase 4" is typically used to describe post-market-approval clinical studies. We are allowing all patients who complete our phase 3 study, to enroll in an extension study in which they remain on the twice-daily DR Cysteamine drug regimen until time of market approval. This study has been reviewed and accepted by FDA. The protocol for this extension study allows for also enrolling patients who did not participate in the phase 3 study. Some patients who were ineligible to participate in the phase 3

based on their medical status at the time (for example patients who have had kidney transplants), may now be eligible to participate in the extension study. Any such patients would be enrolled and monitored through the same clinical sites that conducted our phase 3 study. Currently, only phase 3 patients are participating in this extension study. We will announce to the foundations when new patients will be accepted for enrollment.

## 5 Are there plans to make additional forms of RP 103 for those who are too young to swallow and for those with swallowing difficulties?

RP 103 was formulated specifically with the youngest cystinosis patients in mind. The active ingredient is formulated into microbeads, which have been enteric coated and then encapsulated. The idea is that the capsule can be opened and the microbeads sprinkled onto food or into juice, for example. Raptor is conducting additional clinical studies to determine whether such administration would be viable. It is our hope that cystinosis patients would be put on a twice-daily treatment regimen from the point of diagnosis regardless of their ability to swallow whole capsules.

## 6 Realistically, when do you believe RP 103 will be approved by the FDA? What is the status of the FDA application?

## Q&A with Ted Daley (continued from previous page)

We have a few steps to complete before our documentation will be ready for FDA submission: complete our phase 3 study, capture additional data from our extension study, and other product testing in the laboratory and manufacturing settings. We expect to have the necessary data to complete and submit our New Drug Application (NDA) before the end of 2011. From the point of submission, the FDA's timeline to review and decide on our application will depend on their assessment of the "unmet need" in this patient population. In a case like this where there are existing therapies, a new treatment may be considered as filling an unmet need if its effectiveness is at least equivalent to the accepted treatment, avoids serious side effects of an available treatment, or decreases a clinically significant level of toxicity of an accepted treatment. If the agency believes there is sufficient unmet need, there are provisions in the regulations that allow for a six-month NDA review timeline under "priority review". Otherwise, FDA guidelines are for 10-month review of NDAs, although it is possible it could take longer.

**7** **Currently, Cystagon® is only available through one pharmacy. Once approved by the FDA, will the drug be available to patients at more than one pharmacy?**

We are still evaluating different options for distributing DR Cysteamine once approved. We are committed to making it available to all patients and to making it as convenient as possible to obtain the drug.

**8** **What will be the cost of the new medication? Will insurance companies be likely to pay for the new drug?**

The investments required to develop medications to treat very rare diseases and to bring them to market are substantial. I can't say yet what the price of DR Cysteamine will be assuming it is approved, but it will be considerably higher than the cost of the currently available treatment. We are confident that payers will cover the drug based on expected advantages over the current standard of care. We will ensure that patients have access to the drug regardless of their ability to pay.

**9** **There are patients all over the world with cystinosis. Once the drug is approved by the FDA, how soon will cystinosis patients in other countries receive the drug?**

Decisions on marketing approval in countries other than the US would come after any decision on our FDA submission here. Our goal is to make DR Cysteamine available to cystinosis patients anywhere in the world, whether through registration or special-access programs.

**10** **The fact that cysteamine could benefit other diseases is remarkable. Raptor has the exclusive, worldwide license for EC Cysteamine and has conducted other clinical studies using cysteamine. What other clinical trials are currently taking place using cysteamine? (NASH, Huntington's)? What is the status of those clinical trials and have there been promising results for those patient groups?**

Raptor collaborated with University of California, San Diego (UCSD) on a phase 2a clinical study of DR Cysteamine in non-alcoholic steatohepatitis (NASH). This study was sponsored by the UCSD investigators, and results were reported at the Digestive Disease Week (DDW) conference in New Orleans in May, 2010. Based on the positive findings of the phase 2a study, we have drafted a protocol for a phase 2b study, and we are very excited about the prospects of DR Cysteamine as a potential treatment in this widespread metabolic disease.

Raptor is also collaborating with CHU d'Angers, in France, on a phase 2 study of DR Cysteamine in Huntington's Disease, a genetic neurological disorder. This study began enrolling patients this past fall.

**11** **The Cystinosis Research Foundation is committed to ensuring the delayed-release medication is FDA approved. Practically speaking, what can we do to assist Raptor in their quest for FDA approval of RP 103?**

Raptor is committed to helping people with cystinosis. The collaboration between the Foundation and our clinical researchers has been valuable. One of the greatest challenges in developing therapies for rare diseases is identifying patients to participate in clinical studies. The Foundation was able to help quickly identify patients and investigators, shortening the anticipated time to fully enroll the study. This means that we can potentially bring a valuable therapeutic to you much sooner.



We look forward to continued collaboration, especially on the international patient registry, additional clinical studies, scientific meetings, and other outreach programs, to further our mission of bringing meaningful relief to cystinosis patients and caregivers.

# CCIR Goes Live

**B**etty L. Cabrera, M.P.H. is a researcher at the University of California, San Diego and Curator of the newly launched Cure Cystinosis International Registry (CCIR). She would like to share with you the importance of this patient registry and personally invite you to join.

## *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 such as cystinosis can often be 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 to fight a disease. However, the research community desperately needs the right tool that will permit access to these resources.

*As a scientist, the registry is a unique and critically important resource especially now.*

*Patrick Harrison, PhD, Head, Department of Physiology, University College Cork, Ireland*

A patient registry is a tool widely used 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 only international patient registry for cystinosis, Cure Cystinosis International Registry. The express purpose of CCIR is to make information available to the research community and thus promote accelerated research in advanced treatments and ultimately a cure for cystinosis.



*Betty L. Cabrera,  
CCIR Curator*

## *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 is a top priority. Registered

identifying information is protected using three layers of security: encryption of all internet transmissions, limited and password protected access, and de-identification or removal of all identifying information from communications and reports. Contact with registrants is limited to reminders to update profiles annually, and to notifications of clinical trial opportunities.

CCIR has a designated Curator to oversee the daily operations of the registry. The Curator is available to provide assistance at any step of the registration process, including helping registrants collect clinical information requested in the survey. Once a profile is entered, the Curator reviews it to ensure that the information provided is consistent and that the survey is complete. The Curator also acts as a gatekeeper of the profiles contained in the registry, ensuring that confidentiality is maintained. All requests submitted by professionals asking for data reports or for the delivery of notifications to registrants about opportunities to participate in a clinical trial are carefully processed by the Curator.

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

*Continued on next page*

## Children with Rare Diseases

Children with rare diseases are particularly disadvantaged. Their conditions are often poorly understood by most physicians and fall off the radar at federal research granting agencies that power the progress on “more important” diseases.

Indeed, even an interested investigator or pharmaceutical company with a new idea, finds it a daunting task just to determine how many individuals might be assembled to develop a new diagnostic procedure or test a new therapy.

Fortunately, in this new digital age, people with rare diseases now have the capacity to find each other and speak with one voice to encourage research by participating in disease-specific patient registries.

With the launch of the Cure Cystinosis International Registry in August 2010, the cystinosis community has acquired a real opportunity to press their cause. This web-based registry allows every patient with cystinosis to submit their own experience in privacy and gain access to anonymous information for the whole group. Their physicians can register through a professional portal, providing them with a wealth of practical clinical information about cystinosis based on the anonymous data that accumulates.

**I have no doubt that this new forum will stimulate research on cystinosis in the future and I encourage every cystinosis family to join the effort.**

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

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

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

When you register with CCIR, you are uniting with hundreds of people around the world to combat cystinosis, and 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 cystinosis community overall has responded to the survey questions. You may be interested in checking the results frequently as more people register.

Through the *Ask An Expert* feature available exclusively to registrants, you will have the opportunity to submit questions about the disease that you would like a cystinosis medical expert's response. We are finding that many people share similar concerns and soon a list of *Frequently Asked Questions* and their answers will be available on the website. We have also noted that the cystinosis community has questions, the answers to which are unavailable, or for which little information exists in the medical literature. Survey questions are beginning to capture this information, so we know we are on the right track.

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.

**Age at Diagnosis**



»» *The registry 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

**CCIR progress and goals**

In six short months, 227 people from 28 different countries have registered with CCIR. The majority of registrants are from North America (56 percent) and Europe (24 percent), though the registry has reached cystinosis patients as far as South Africa and Russia. In an effort to reach more people the registry is already available in Spanish, and we will have French and Portuguese translations available in the near future.

We need your help to build this invaluable resource, which could lead to improvements in the lives of cystinosis sufferers everywhere. The time is now! What are you waiting for? If you have already joined, thank you, and please don't forget to spread the word.

We encourage you to post a link to CCIR (<http://cystinosisregistry.org>) on your Facebook page or contact Betty Cabrera directly with any questions you have ([curator@cystinosisregistry.org](mailto:curator@cystinosisregistry.org)).

**CCIR PARTNERS**



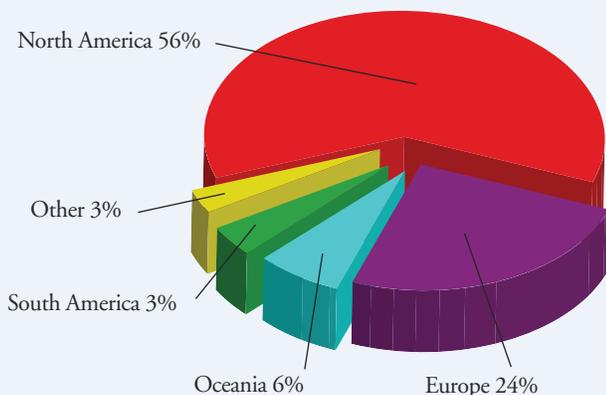
**CCIR ADVOCATES**



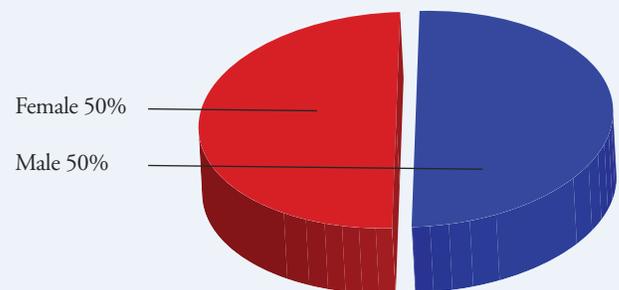
**The Cystinosis Foundation**

Cystinosis Foundation New Jersey Chapter  
Cystinose France

**Geographical Location of Registrants**



**Gender of Registrants**



The three charts above are based on current demographics from the CCIR. Registrants have ongoing access to CCIR data.

## There's Solace in Numbers in the World of Rare Diseases

In the world of rare diseases, the story of the Daniels family in Kraaifontein, Cape Town, is probably typical.

For instance, it took parents Gail and Colin Daniels more than 11 years to put a name to the condition, cystinosis, that had dogged their daughter, Danielle, since birth. The disease has stunted her growth, slowed her learning, made her a fixture in doctors' offices and hospital wards and subjected her to a rigid diet of supplements and boosters.

The family had its first scare when Danielle was hospitalized when she was only a few months old. By age two, doctors looked at her small frame and bowed legs. She "waddled," her mother said. She suspected that her child had rickets, a disease, common in Africa, that is characterized by the softening of what are meant to be growing bones. It is caused by a shortage of both vitamin D and calcium.

### **Narrowing Down the Condition**

But after months of tests, mad dashes to the hospital, a string of specialists and a babble of misdiagnoses, doctors finally narrowed down her condition to Fanconi syndrome.

Named after a Swiss paediatrician, the syndrome affects the ducts or tubes leading from the kidney. These ducts are meant to salvage essential nutrients from fluids passing through the kidneys. But in this syndrome the ducts are unable to do so and precious glucose, carbohydrates, phosphates, amino acids and minerals are passed out in the urine. Without those essential nutrients a rogue's gallery of troubles follow, rickets and weak bone formation – leading to that waddling and underdevelopment – among them.

There is no cure for Fanconi. Patients are put on a lifelong course of boosters, supplements, vitamins, phosphate replacements and citrates to make up for the shortfall of minerals.

So for the years that followed Danielle's life revolved around daily handfuls of pills, potions and powders. She dared not skip a dosage. "It's normal now," she says of this regimen.

Despite the medication, her problems persisted. There were threats of kidney failure – she was in and out of hospital and her eyesight weakened enough to scare the family. Also patients are commonly lethargic, so learning is slow.

Her mother, a teacher, became increasingly frustrated with evasive doctors. So she started her own research, foraging whatever she could from newspapers and the web. "The funny thing was that I couldn't tell the doctors anything I knew," she says. "They didn't want me to know."

But it was this research that pointed her in the direction of cystinosis. "The name kept coming up whenever I read about Fanconi."

### **Naming the Disease**

Cystinosis borrows its name from a namesake gene, the cystinosis gene. Also known as CTNS, the gene provides the instructions to produce a transport protein – cystinosin – inside compartments in the cell known as lysosomes.

Inside the lysosomes, cystinosin is broken down into its smaller building blocks, amino acids, including one known as cystine. Living up to its job title, the cystinosin then carries the cystine out of the lysosomes.

When it is unable to do so, the cells produce too little cystinosin or the cystinosin is too weak to carry out its transport duties. Cystine accumulates in the lysosomes and grows into crystals. These crystals then destroy the lysosomes, the cells and ultimately the body's tissues.

The crystals typically form in the brain, liver, thyroid, pancreas, muscles and particularly in the kidneys and eyes. In the kidneys it damages ducts that can lead to full-blown kidney failure,

necessitating kidney transplants. In the eyes the cystine crystals build up in the cornea, triggering extreme sensitivity to light and eventual blindness.

Cystinosis can be put down to a stroke of poor genetic luck. Both parents, unaffected by the condition, have to carry the faulty CTNS gene. Each parent then passes on a copy of that gene to their child.

Statistically, there's a 25% chance that parents carrying the mutated gene will have a child with cystinosis. Gail and Colin have a second child, a son, Dane (20), who does not have cystinosis.

A childhood disease, cystinosis is considered rare. It is said to affect one child in every 100,000 to 200,000 live births. According to available statistics, only about 2,000 people around the world have been diagnosed with cystinosis. But elementary mathematics – one in 200,000 in a world population of 6 or 7 billion people – would suggest that the number is way off the mark.

"A lot of children probably die from this condition without being diagnosed, especially in the developing world," said Dr. Peter Nourse, the paediatric nephrologist at Cape Town's Red Cross Children's Hospital, who treats Danielle.

With about 14 patients at Cape Town's Red Cross and Tygerberg hospitals, Nourse has good reason to think that the disease is more common than statistics bear out. "We haven't worked out the true incidence in South Africa, but it does seem that there are quite a lot of cases," he says.

### **Treatment**

Although there is no cure for cystinosis, there is a treatment in a drug called cysteamine. It was Gail who stumbled on it in a newspaper article about another Red Cross doctor who was treating cases of photophobia with eye drops he'd cooked up, using cysteamine as the main ingredient.

Cysteamine breaks down the stored-up cystine into a similarly named amino acid, cysteine, and a cysteine-cysteamine mix, both of which are flushed out of the lysosomes. But one needs regular fixes: a cysteamine drink (drug plus distilled water) has to be downed every six hours like clockwork, while eye drops are administered hourly.

Also, the cocktail tastes “disgusting,” so doctors, parents and caregivers have their work cut out feeding it to children four times a day, says Nourse. Even hardened nurses have been known to recoil at the smell.

Danielle is more fortunate. She takes an easier-to-keep-down capsule version of cysteamine, Cystagon®, which costs about R18 000 a month, a bill footed by the family’s medical aid scheme.

Cystagon® was developed as an orphan drug – medication for rare diseases – in the United States, where it has been prescribed for cystinosis for the past 15 years. It’s not readily available or licensed in South Africa, though. So the Daniels family also had to jump through a number of bureaucratic hoops to have the drug imported.

There’s the potential of an even sweeter pill on the horizon. Raptor Pharmaceuticals in the US is making headway on a delayed-release version of the treatment, to be known as DR Cysteamine. It’s hoped that DR Cysteamine will reduce some of Cystagon’s side effects and halve the daily dosage.

“The new formulation should be better tolerated and taken only every 12 hours,” reports Margret Kamel, a research coordinator with a team conducting phase-three human trials on the drug at Emory Children’s Centre in the U.S.

### Joining Forces

But how is it that cystinosis is worthy of all this attention? After all, the markets for treatments for most rare diseases are usually too small to coax companies to do the necessary R&D.

“Funding for rare diseases is very uneven,” points out cystinosis guru Dr. William A. Gahl, clinical director at the National Human Genome Research Institute in Maryland in the U.S.

“Cystinosis, too, languished for a while.”

But the condition piqued academic interest because of its “intriguing basis” as a lysosomal transport defect, says Gahl. Researchers hope that treatments for a few dozen inherited metabolic disorders, also linked to faulty lysosomes, can be developed on the back of cystinosis studies.

Cystinosis also benefits from the American Orphan Drug Act, which allows pharmaceutical companies to sell their orphan drugs without fear of competition for a full seven years.

### Families Push for Research

Most of the credit should go to one American family. Under the banner of the Cystinosis Research Foundation (CRF) they established in 2003, Nancy and Geoffrey Stack, whose 19-year-old daughter Natalie has cystinosis, have raised around \$12 million, money they distribute in research grants. That makes the family the largest supporter of cystinosis research in the world.

It was a CRF-funded researcher, for example, who did the groundwork for the new delayed-release cysteamine. The foundation’s latest project is the establishment of the first and only international patient registry, named the Cure Cystinosis International Registry.

“Our goal is to get every patient with cystinosis to register,” says Nancy. “By doing so, we’ll have a more thorough picture of cystinosis and its complications, providing our research teams with information they so desperately need to discover novel treatments and a cure.”

And a cure is exactly where the CRF is headed next. Recently the foundation established the CRF Gene Therapy Consortium, which includes experts in gene and stem-cell therapy.

Working with mouse models, researchers at Scripps Institute in San Diego have used stem cells extracted from bone marrow to express a more shipshape CTNS gene. This revitalized gene was then able to reverse the accumulation of cystine in the cell and subsequent cell death.

The scientists are optimistic but cautious. “These data are very promising,

but the translation from mice to humans does not always reproduce the same effect,” says Dr Stephanie Cherqui, assistant professor in the department of molecular and experimental medicine at Scripps.

### Support and Education

The Daniels family is playing its part. In 2010 Gail and Colin established the South African Cystinosis Support Group, affiliated to the U.S.-based Cystinosis Foundation, which is distinct from the Stacks’ Cystinosis Research Foundation, but the groups work hand in hand on, for example, the international registry.

Gail is mother hen to others in the South African group, largely state patients with no medical aid. She’s produced easy-to-follow information pamphlets for the group and meets and advises members. Until recently Colin covered most of the running costs and lent other assistance, such as cooler bags to preserve the cysteamine drink. (The group now receives a small donation from a Dutch couple, Jan and Marjolein Bos, who run a similar group in the Netherlands.)

Gail wants group members to have the same kind of support she received through the Cystinosis Foundation. “The foundation was the best thing that could have happened to us,” she says, “because we could speak to people who understood what we were saying and what we had gone through.”

Her mentor on the foundation is the eighty-something founder, Jean Hobbs-Hotz. “There is a treatment for cystinosis,” says Hobbs-Hotz. “We want the medical community to understand the disease so they can make a proper diagnosis and for families to understand the disease and the need for compliance.”

Danielle remains on the verge of kidney failure, but her family is keeping their eye on the future. “To say the worst is over, isn’t very realistic,” says Gail. “But we are hopeful and prayerful that by God’s grace a cure for cystinosis will be found so that Danielle and many other sufferers will be able to enjoy good health.”



*The five Helbling children who were diagnosed with cystinosis in the 1950s and 1960s.*

## Much Has Changed in 43 Years

By Carmella Helbling

**M**y husband, Bill, and I were blessed with ten children. We lost five of them in childhood to a mysterious kidney disease – what we now know as cystinosis – before the causes, effects and treatments were well known. None of our other five children had the disease, and all grew to become healthy adults, becoming parents (and in one case, a grandparent) themselves.

Looking back now, we realize how unusual our situation was. There were no support groups in the 1950s and early 1960s. Kidney disease was not nearly as well understood as it is today. Physicians in rural northern Idaho did their best but weren't able to offer much help. We lost two children before we even went to the University of Washington Medical Center in Seattle. Here are their stories:

David was born Jan. 18, 1957. At this time, they did not put children on dialysis. The doctors described his condition as “renal insufficiency with renal rickets.” He was put on citrate solution, extra calcium and other medicines. He got the mumps in June of 1963 and shortly after, passed away at home, July 11, 1963, age 6½.

In 1958, we took David, to Dr. William Mannschreck, pediatrician in Lewiston, ID. He took care of all our cystinosis children. He was a very dedicated doctor and did so much for us.

Steven was born May 20, 1958. The following year, doctors suspected he had the same disease. They started testing, but he became ill when he was just 19 months old, and passed away Dec. 21, 1959.

John was born May 26, 1960. Doctors started tests at an early age and diagnosed him with cystinosis. We began going to Seattle to see Dr. C. Patrick Mahoney, a pediatrician, who had experience with other cystinosis patients. When John was 8, Dr. Malcolm Holliday of San Francisco began looking for a kidney donor for him. On May 9, 1968, John received a kidney transplant at the University of California Medical Center – what may have been the first transplant from a cadaver to a cystinosis patient. (Before then, a kidney would have been taken from one of the parents.) After his transplant, John remained in California; he returned

to the hospital in early July and passed away July 19, 1968.

Brian was born March 4, 1968, just before John went to California. When Brian was about 6 months old we took him to the University of Washington Hospital, for a week of tests. Unfortunately, when he was 5½, he got very sick on Christmas Day and passed away suddenly the next day, Dec. 26, 1973.

Our daughter Mary Ellen was born Sept. 24, 1965. She had a stronger and longer childhood than the boys –

I HOPE THAT BY SHARING OUR STORY, OTHER PARENTS WILL KNOW THAT IT'S POSSIBLE FOR A FAMILY TO GROW STRONGER AFTER ENDURING THE CHALLENGES OF HAVING A CHILD WITH CYSTINOSIS.

attending school and even learning to ride a bicycle. Just before her seventh birthday, we learned that doctors had found a kidney for her. She received the transplant at the University of Washington Hospital, but had complications, eventually rejecting the kidney after a few months.

Next, Mary Ellen started dialysis at the UW and continued it at Children's Hospital in Seattle. Meanwhile, Sacred Heart Hospital in Spokane had a dialysis machine that they no longer needed, so they gave it to us to use. My husband, Bill, and I learned to operate the machine at the Northwest Kidney Center in Seattle. We were the only ones in Moscow who knew how to run the machine, as none of the doctors in the area were familiar with it. We even had to install a reverse-osmosis water purifying system to use with the kidney machine.

Mary Ellen was on the machine for 12 hours, three times a week. It took 21 hours to prepare for the next dialysis because the water had to be heated to 400 degrees to purify it and then cooled down to body temperature. In between dialysis sessions, Mary Ellen went to school but sometimes she was very tired. She was on dialysis for 2½ years before she received another transplant on

with our big family. There was hardly any information about such a rare disease; even the doctors were unfamiliar with it. I don't think we met anyone who had a kidney machine in their home like we did. Those in Seattle area went to the dialysis center, so we really didn't have contact with other families once we got home to Moscow.

Our sick children couldn't do what other children did. They would look out the window and only wish. And we wished for them, but there was not much we could do about it. Our five other children of varying ages had a different upbringing. We couldn't do many of the same things that other families could, such as long vacations, camping or weekends at the lake. I think this made our surviving children much more considerate and respectful of others – it definitely made them better people. We are proud of our sons, Mike, Jeff and Bill, and our daughters, Barb and Karen. They have blessed us with 13 cystinosis-free grandchildren and one great-grandson.

Today, groups like the Cystinosis Research Foundation are developing new treatments and trying to find a cure. New peritoneal dialysis machines are so much smaller and better than the one we used in the 1970s. At the dialysis



*Mary Ellen Helbling and her mother, shown in the hospital and with the donated dialysis machine in the family's home.*

Dec. 15, 1975, when she was 10 years old. She came home to Moscow on Feb. 19, 1976, continued her dialysis and went back to school. She had a good month, but on March 16, she became ill after coming off the machine. We rushed her to St. Joseph's Hospital in Lewiston, where she passed away March 17, 1976. We still hear from one of Mary Ellen's nurses more than 30 years later.

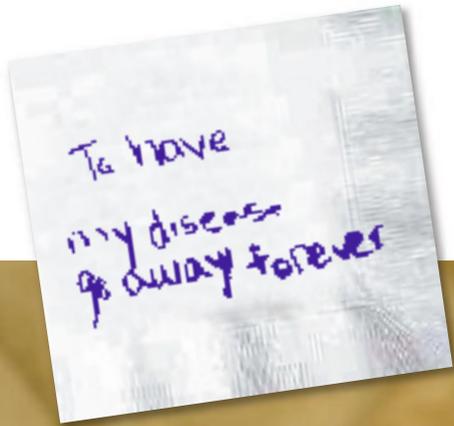
Our faith in God probably is the only thing that kept us going during our children's struggles with this disease. Now, looking back, I don't know how we ever managed

center in Moscow, a nurse showed me the latest peritoneal dialysis machine. It was simply a big box, which she carried with no problem – a lot different from the big stainless steel machine we had at home. I couldn't believe it.

We're hopeful that researchers will continue to make improvements in care and treatment, as well provide support for families with children who develop the disease. I hope that by sharing our story, other parents will know that it's possible for a family to grow stronger after enduring the challenges of having a child with cystinosis.

*Carmella Helbling lives with her husband, Bill, in Moscow, Idaho, where they support cystinosis research through contributions to local fundraisers. She is a resource to other parents in Idaho and Washington. She may be reached at [bhelbling@moscow.com](mailto:bhelbling@moscow.com)*

**A Hopeful Note from a Child's Heart Sparks  
a Foundation of Possibility for Those with Cystinosis**



*Guided by a Wish.*

By Dennis Arp

*I*t started with seven words scribbled in purple crayon on a paper napkin, folded and stuffed deep into a pocket. A dream, perhaps too big and too bold for a child to believe it deserved to be dreamed at all.

*“to have my disease go away forever.”*

Natalie Stack was a day away from her twelfth birthday when her mom asked over lunch at a local restaurant what her celebratory wish might be. Tears filled Nancy Stack’s eyes as she held her daughter’s most cherished desire tightly in her hand.

“It’s OK, Mom,” Natalie whispered. “I’ll be all right.”

Now, nearly a decade later, Natalie’s wish has never been closer to reality. Thanks to the foundation she inspired, Natalie and the 500 or so other cystinosis sufferers across the nation (about 2,000 worldwide) are gaining relief from the daily, wrenching effects of this incurable disease, which disproportionately attacks the young. They can indeed see hope on the horizon as research yields breakthroughs that open new worlds of opportunity for better treatments and perhaps even a cure.

The impact of the Cystinosis Research Foundation is front and center – in laboratories and drug trial centers, in board rooms and living rooms, in hospitals and homes. But that impact is never more dynamic or immediate than when it transforms the dreams of cystinosis sufferers themselves.



*Natalie and  
Maison in 2003.*

*Continued on next page*

“Natalie’s wish was the catalyst in our quest to find a cure,” says Nancy Stack, CRF co-founder with her husband, Jeff, and chair of its Board of Trustees, recalling the day in 2003 when she persuaded her daughter to share her birthday wish. “Those seven words motivated us to establish the foundation. We knew at that moment that we needed to make every effort to realize Natalie’s wish, and the wishes of others with cystinosis.”

In many ways, the CRF’s achievements so far provide a blueprint for how an indomitable commitment can break through barriers that typically block research progress on rare diseases. The successes also illuminate the ways love and friendship, science and support can turn a bleak diagnosis into a beacon of hope.

That hope was in short supply the day 19 years ago when Nancy and Jeff Stack first learned Natalie had been diagnosed with cystinosis.

“In a single minute, the life we envisioned for Natalie and our family changed dramatically,” Nancy recalls.

They stared at a future dominated by a metabolic disorder that was causing the amino acid called cystine to accumulate in her cells. There was no cure, and Natalie needed to begin taking a drug with a horrible taste

Oh, and the course of therapy required dosing every six hours, which meant she had to be awakened in the middle of the night, sometimes having to be held down to endure the unpalatable treatment.

For hundreds of cystinosis families, the stresses and strains of such treatment are all too familiar. At the same time, the daily trials galvanized the family resolve shared by Nancy, Jeff, Natalie and her sister,

incentive to make and market new medications, given the relatively small number of those afflicted.

What’s more, with the development of cysteamine as a treatment for cystinosis, affected children started living enough beyond infancy, so the inspiration for new research seemed to go away.

“Our children stopped dying before they turned 10, and that was wonderful,” Nancy Stack says.



Alex. The four also drew strength from the support of other family members as well as old friends and new ones they met by reaching out to the cystinosis community.

“But we knew that wasn’t enough. We knew we needed to push for more information and fund more research.”

The CRF was launched in 2003 with a reception in Orange County attended by about 90 of the Stack’s friends. A physician explained the devastating realities of cystinosis, and by the end of the evening, more than \$180,000 had been raised to seed the new research foundation.

That first group of contributors embraced Natalie’s wish and made it their own, sharing the stories of hope and recruiting others to the cause. One key to the foundation’s fundraising success is the knowledge that 100 percent of donations go to finding better treatments and ultimately a cure.

“Our children stopped dying before they turned 10, and that was wonderful,” Nancy Stack says. “But we knew that wasn’t enough – that we needed to push for more information and fund more research.”

and smell just to slow the destruction of her liver, kidneys, eyes, brain and every other organ in her body. Plus, the daily regimen of medicine would bring its own complications, requiring her to take four or five other medications to control the side effects.

Wanting to do more to make progress toward a cure, the Stacks dived into their own research, learning that cystinosis is an “orphan disease” – a disorder difficult for the pharmaceutical industry to embrace. Such a disease provides little financial

The momentum that began with Natalie's wish and grew with the success of the foundation she inspired has grown so strong that families around the world are raising funds for the CRF.

And the impact continues to grow. After its first call for grant proposals in 2004, the foundation received five responses from research institutions. Now the CRF announces research calls twice a year and has received as many as 15 proposals for a single call.

"It's rewarding to know that with the help of our supporters, we've been able to bring new researchers into the field," Nancy says.

Most recently, the work of Stephanie Cherqui, PhD, and her team at The Scripps Research Institute in La Jolla, California, has yielded a plan with exciting potential. Though many hurdles and more years of research remain, this stem-cell therapy holds great promise as a path to a cure.

Throughout the evaluation of all of CRF's proposals and laboratory research there has been the tireless work and steadfast leadership of Jerry Schneider, MD. A pioneer in cystinosis research, which he began in 1965, Dr. Schneider served as chair of the CRF Scientific Research Board, recruiting, training and mentoring many in the field whose work is now bringing us closer to a cure.

*Continued on next page*



**Thanks to the generous support of countless contributors, the Cystinosis Research Foundation is helping to make a significant and difference in the lives of those with cystinosis:**

- A new medication is in Phase 3 and Phase 4 trials, with early 2012 approval by the FDA a genuine possibility. Delayed-Release Cysteamine, will mean fewer doses and fewer side effects, fueling a great leap in quality of life. For instance, those with cystinosis will enjoy the simple pleasure of sleeping through the night for the first time in their lives.
- Researchers are also making progress in a quest for the cure. Extraordinary discoveries using gene and stem-cell therapy have reversed cystinosis in laboratory mice and hold the potential for advances combating a range of genetic disorders. And now CRF's Cystinosis Gene Therapy Consortium is bringing together top leaders in the field for the sole purpose of developing breakthrough treatments targeting this crippling disease.
- The CRF has established the only cystinosis patient registry, making it faster and easier to involve patients in clinical trials. The registry also gives researchers and physicians, for the first time, a collective window to symptoms and other issues experienced by patients. This information is crucial to developing treatment plans or considering the potential for new areas of research.
- Since 2003, the CRF has funded 78 studies and fellowships, with 100 percent of the nearly \$15 million raised committed to this critical research.

**These accomplishments and more were mere pipe dreams a decade ago, when those with cystinosis and their families had little reason to dream beyond the next treatment.**



*The delayed-release medication has changed my life,” Natalie says. “I hope it will soon be available for everyone who has cystinosis.”*

With CRF funding, Dr. Schneider and Dr. Ranjan Dohil at UC San Diego developed the delayed-release form of cysteamine that has been licensed to Raptor Pharmaceuticals. When EC Cysteamine gains FDA approval, hopefully in 2012, cystinosis patients will be able to take their life-extending medication every 12 hours instead of every six – a tremendous boon for quality of life.

“Your perseverance gave us hope,” Nancy and Jeff Stack wrote in a thank-you letter to Dr. Schneider, whose work was celebrated at a retirement dinner in 2010. “We are closer to a cure than ever before, and we have you to thank for that.”

Natalie Stack shared her own message at the tribute event.

“Dr. Schneider, you saved my life,” she said. “Without you, I would not have lived this long.”

And what of Natalie’s life these days? Well, thanks to advances in treatment, the 20-year-old isn’t just getting by, she’s thriving.

A year ago, she transferred from Loyola Marymount University in Los Angeles to Georgetown University in Washington, DC, and reports, “I love the people, the community and the challenges. It’s a perfect fit for me!”

Being a part of the EC Cysteamine trial has allowed her to live far from her home in

California. She still has to take eight different medications each day, but because the dosage is only every 12 hours, the burden is less onerous and the side effects are reduced.

“The delayed-release medication has changed my life,” she says. “I hope it will soon be available for everyone who has cystinosis.”

Hope has never been a more vibrant part of Natalie’s life. It’s with her as she heads to her favorite classes in psychology, sociology and French, as she meets new friends and explores

new surroundings, as she plans her next trip home to see her family, including her cat, Max, and her puppy, Dillon.

She knows that the challenges of cystinosis are as close as the next blood draw, the next dosing, the next doctor visit. But her days are also energized by planning for a future full of possibility.

Days as bright as a child’s wish captured in purple crayon:

“To have my disease go away forever.”

A dream too big? Not anymore.

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*Dennis Arp is a freelance writer and author based in Southern California.*



*Natalie with her sister Alex.*

# Hope in a Life Filled with Hurdles

**I have lived with cystinosis for 32 years and it seems like everyday  
I face another hurdle — each one bigger than the previous one.**

I currently face possibly my most challenging and scariest hurdle yet, a kidney transplant. For years it seemed like I was prolonging the inevitable, and in many ways that is exactly what I have done. I am very lucky, blessed, and thankful that I have been able to avoid such a major hurdle for so many years. It is almost unheard of for a cystinosis patient to have their own kidneys for such a long time. 32 years seems like forever for someone who wasn't supposed to live past age 10.

When I was diagnosed with cystinosis in 1980, my parents were told that I would grow to only three feet tall; that I would never go to school; and that I would need a kidney transplant by age 9. I have overcome many odds, including reaching the height of five feet, two inches, earning a master's degree in Speech Language Pathology (and currently working on an RN degree), and not needing a kidney transplant until I was 32 years old.

The transplant process is long and slow and I have barely begun. Florida Hospital recently approved me as a "suitable recipient," but today, when good kidneys are few and far between, they won't just give a kidney to anyone. I am beginning donor testing procedures in which every potential donor must willingly volunteer to go through a rigorous regimen with numerous medical tests.

It is a scary and overwhelming time for me and my family. Time is of the essence since I only have 13–15 percent kidney function left and I am doing everything possible to avoid dialysis at all costs. I will require dialysis when my kidney function reaches 10 percent. My dad and my sister are prime donor candidates. My nephrologist says that a kidney from a living donor is better than one from

a deceased donor; a family member's kidney is better than a non-family member's; a male kidney is more desirable than a female kidney; and a younger kidney is better than an older kidney. By "better" I mean a longer-lasting kidney. The last thing I want is multiple kidney transplants. I want one kidney and then I want to be cured of cystinosis forever.

The average life of a kidney from a living relative is 17 years. This number fluctuates based on the donated kidney. The million-dollar question yet to be answered: do I take my father's kidney or my sister's kidney? My nephrologist believes that I should take my dad's, even though he is 63 – a relatively "old" kidney – while my surgeon believes I should take my sister's because she is much younger. My nephrologist thinks I should "save" my sister's kidney in case, God forbid, I should need another one in the future. My dad has already begun testing.

Although cystinosis has placed many roadblocks in my path, I have overcome each of them. I know one day I will look back and realize this was just another obstacle that taught me strength and resilience. God has brought me through every challenge I faced and I believe my kidney transplant will be no different.

Cystinosis has made me the person I am and for that I am thankful. I continue to dream and hope and know that when all is said and done I will come out on top in spite of all I have had to deal with.

Thank you CRF for your dedication to the cystinosis community. Through your research and funding I am able to dream about a life where I no longer live with cystinosis!



*Whitney with her dog Tasha.*

**Whitney Glaze**

# Addison Rose: A Fighter with a Smile

By Nicole Cox, Addison's mom

Addison Rose Cox was born on October 5, 2008. She was a healthy baby girl weighing 7 pounds 10 ounces. Her first year was filled with so much fun and laughter. We watched in awe as she grew into a little human being with her own personality.

When Addison stopped nursing around 10 months, she didn't eat very much. I would watch in amazement as the other children ate everything that was put on their plates. We never thought anything about it. We just thought we had a picky eater. And our pediatrician assured us that it was normal for children to go through stages of not eating.

Addison would go three or four days without eating a thing. That's when we knew something was wrong. Still, no one would believe us. At Addison's 15-month check-up, she fell off the weight charts and

Our nephrologist wanted to run one more test, to rule out all the possibilities. I remember asking him what it was for, and he told me that it was for a disease so rare, he wasn't even going to tell us about it because it would scare us.

In April we finally got the news that Addison had cystinosis. I will never forget the doctor's words, "You have won the reverse lottery." Like most people, we had no idea what cystinosis was. I am sure that the doctor explained it all to us, but I don't remember a thing. All I heard was, "Your daughter is very sick and there is no cure." When we left the doctor's office that day, we had no idea how much cystinosis would change our lives.

Addison was started on Cystagon®, along with the slew of other medicines she was already taking. She took the medicines like a champ! We were amazed at how brave and resilient our little girl was. But before long she

began to fight us and throw up the Cystagon®. The nights were the worst. I hated waking her up to force medicines down her throat. Not to mention changing the sheets because her bed was soaked in urine.

I remember one particular night, sitting on top of Addie, holding her arms and legs, trying to give her Cystagon® for the fourth time. We were all crying. I looked up at Brandon and said, "I can't do this anymore."



We made an appointment with a surgeon the next day to discuss a g-tube. When we finally scheduled her surgery, Addison weighed barely 20 pounds and still losing weight. We no longer had a choice, she now needed the surgery to save her life. Since her g-tube surgery in October, Addison has gained five pounds and her energy has increased exponentially. It is such a blessing to know that she's getting enough calories and all her medicines.

On the outside, she is a normal two-year-old. No one would guess she is so sick. Her personality just bursts out of her little body. She loves to dance and sing and has never met a person she didn't like. She enjoys going to school, playing soccer and hanging with friends.

But more than anything she is a fighter. With all that she has gone through, she always has a smile on her face. She is so courageous and she has taught me so much this year.

Less than a year after Addison's diagnosis, we are so hopeful about the life Addison has ahead of her. The Cystinosis Research Foundation has been an amazing blessing. With the delayed-release drug and all the research being funded by the CRF, we believe that we will see a cure in Addison's lifetime.

**Thank you, for giving Addison and all those with cystinosis hope.**



Addison with her dad Brandon, her mom Nicole and her baby sister Emma.

red flags immediately went up with the doctors. We were sent for the first of many blood work ups.

Over the next three months we went through a number of diagnoses. First, we were told that Addison had renal tubular acidosis. After more blood work and a bone x-ray, we were told that she also had rickets.

# Holt Grier: A Zest for Life

Five-year-old Holt continues to thrive and inspire us.

He began his final year of preschool in August after having a big summer with his brother and sister, aunts, uncles, cousins, grandparents and friends. Whether swimming at the pool, playing with his neighborhood friends or enjoying family vacation (yes, he got to go back to his “Bahamas House”) he lives every day to its fullest and is never without a smile, a hug and enthusiasm for being in the moment. He is truly an inspiration to us every day.

Despite his rigorous schedule of six medicines every six hours every day through a g-tube in his stomach, Holt is vibrant, passionate, charming and bursting with a zest for life.

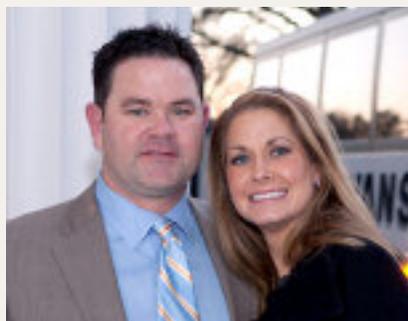
fine motor skills, as well as build his confidence in the classroom. Our entire family is passionate about being part of the Davidson Day family as the students, staff and faculty there have embraced him since day one.

Holt spent a lot of time this fall roaming the football field with the Davidson Day School football team where his Uncle Chad is the head coach. He has enjoyed getting to know the players and has earned a special place in each of their hearts. One of his favorite things to do while at practice was to blow the whistle to



t-ball coaches threw a cornhole tournament over the summer that raised money for cystinosis research. His Aunt Aubrey and Uncle Brandon worked hard planning the fourth annual Hearts for Holt event that took place on February 12, 2011. Like past events, it was a huge success.

We are proud to be partners with the Cystinosis Research Foundation.



Jason and Chrissy Grier (center) at the annual Hearts for Holt celebration. Other photos show a few of the 300 plus guests who attended the 2011 event.

While his energy level can drain quickly due to muscle wasting – he will take a rest when appropriate, grab some water and get right back into the thick of the action. He is enrolled in the Pre-K Montessori class at the Davidson Day School and is able to see his brother and sister and three of his cousins every day. With the help, patience and understanding of both of his teachers, the Montessori approach has been extremely instrumental in helping to further strengthen Holt’s

start a series of drills. Holt is playing basketball this season with the Huntersville Youth Athletic Association. He loves playing defense since this involves standing in front of the person with the ball and waving his arms around. Though he is thoroughly enjoying basketball, Holt is really looking forward to the upcoming t-ball season. Because of his determination on the field last season (and maybe because of his precious, curly hair), one of Holt’s

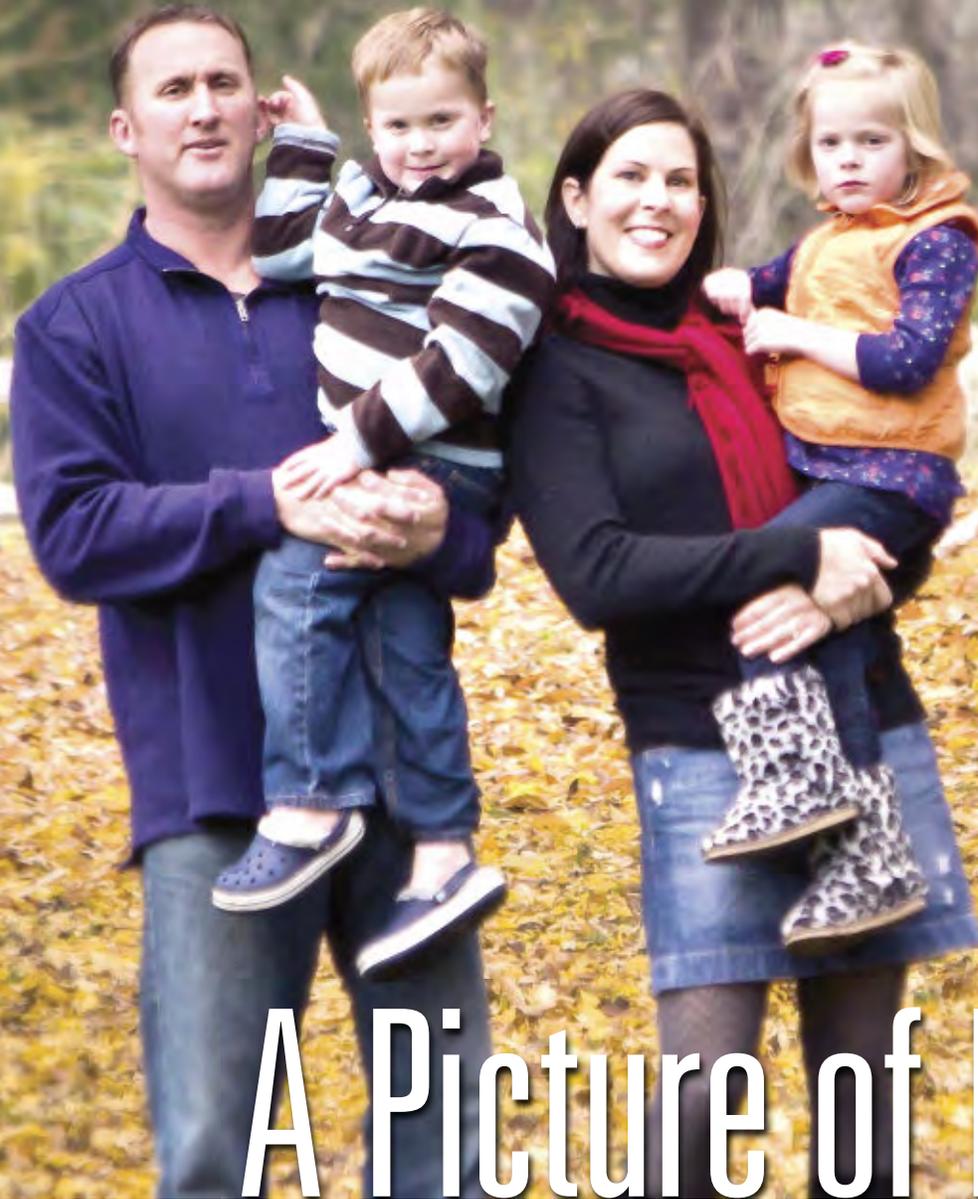
And we are thankful for all of our family and friends who have been so compassionate, understanding and caring throughout the past four years.

We are hopeful for the future and are confident in the promise of the research and the impact that it will have on Holt and so many others.

*Love,  
Jason and Chrissy,  
Mary Logan, Jack and Holt Grier*

Greetings to our family and friends across the United States and beyond! Our children, Jenna and Patrick, are six-year-old twins who were diagnosed with cystinosis at 15 months of age. Our lives are busier and more hectic than ever. While we are still sad and fearful much of the time, today we are more hopeful than we have ever been.

*Patrick and Jenna Partington  
with their mom and dad,  
Teresa and Kevin.*



# A Picture of Hope

Soon after Jenna and Patrick's diagnosis I was asked to write our first CRF newsletter update, and here we are five years later! While the fear and sadness remains from that first year, we now have more hope for our kids' futures than we ever imagined possible. We are filled with gratitude to those who are the Cystinosis Research Foundation. The reports you are reading in this magazine are the words we live for – encouraging words that offer hope in this unpredictable, exhausting and often heart-wrenching journey.

Jenna and Patrick began kindergarten in autumn 2010 and have enjoyed their first months of school. Part of getting our children acclimated to school was helping them – and their teachers, classmates and classmates' families – understand the disease that we deal with day to day. The kids are able to keep water at their desks so that they can satisfy their two-gallon-a-day (each) water cravings.

*I shudder to consider what happens inside Jenna's little body today, knowing that damage control has been suspended and the disease is free to progress in the subtle and insidious ways that it does.*

School staff and other parents have begun to help dispense Cystagon®, keeping the kids on their every-six-hour dosage schedule. The kids continue with their other medications (eight drugs, now including Lipitor, make up their twice-a-day “cocktail”), which they are able to take at home in the morning and evening. Both kids are uncomfortable with stomach and headaches for about an hour after taking their medicine, which often makes starting the school day a struggle. But we continue to be awestruck by their ability to put on a brave face and fight through the rough moments. Jenna and Patrick are happy kids, who love their school friends and are thus far able to enjoy all the activities that their pals enjoy. For this we are so thankful.

Fundraising and awareness efforts in Sacramento have continued as *Jenna & Patrick's Foundation of Hope* hosted *Swing & Bling #2* last October. The full-day event included golf and an evening dinner at Sacramento's beautiful Citizen Hotel. With the help of an amazing group of supporters the foundation raised more than \$260,000. In addition, Holy Spirit School and Giovanni's Pizza, CREW (Commercial Real Estate Women), and Tooley Oil Company hosted an event that raised \$13,000. One hundred percent of these dollars will be passed along to the Cystinosis Research Foundation, where every dollar contributed

is thoughtfully and quickly distributed to doctors and researchers – who are doing the real work of finding improved treatments and, God willing, a cure for cystinosis.

As I write this update Jenna is sick with a flu bug that has her in bed with a fever, sore throat, stomachache and headache. While she fights this common sickness, her body also fights the same fight that it has since she was born. Today she

won't get her Cystagon® every six hours; she won't get her daily medicine cocktails; and she will have trouble keeping down the water that she craves. She will get none of these treatments that keep cystinosis at bay. I shudder to consider what happens inside Jenna's little body today, knowing that damage control has been suspended and the disease is free to progress in the subtle and insidious ways that it does.



As a reader of this magazine, you are a valuable supporter who has undoubtedly made an effort to learn about cystinosis and share stories that create awareness about it. And you have likely contributed your share of dollars to support ongoing research. What a feeling it is to know that our children and so many others are being looked after in such a loving way!

Thank you.

***Teresa, Kevin, Patrick and Jenna Partington***

# How Are the Kids Doing?

Here is my best synopsis regarding the state of Jenna and Patrick, and the state of cystinosis, the genetic disease they both have. My husband, Kevin and I joined 16 people – “friends of the Cystinosis Research Foundation” – recently for roundtable discussions and various presentations about the disease and the children who have it. Anyone who asks, “How are the kids doing?” should know a bit about what was shared and what I learned during our weekend retreat.

## Regarding the “Friends of the CRF”

On February 19 and 20, CRF founders Jeff and Nancy Stack very generously hosted a lovely retreat in San Francisco where a few families, friends, patients and a brilliant doctor from the cystinosis community shared what they know and what they hope will be. It would be impossible to convey the energy in our small conference room, or the camaraderie and friendship shared over dinner and drinks. I was even able to attend a church service with another mother who also has a child with cystinosis.

I will never forget the message at the service, or our walk arm-in-arm as we braved the steep streets of San Francisco in high heels and in the rain.

Attendees that weekend were a small but mighty army gathered in the name of a rare disease. The room was filled with business prowess, medical brilliance, and pure honesty and trust. The passion of every person attending made it abundantly clear that cystinosis doesn’t stand a chance!

## Let’s Not Sugar Coat It

“How are you doing? How are the kids doing?” Before I answer let me say that I am blessed, and I know my journey is meant for me, though I may never completely understand it.

**Here’s how we’re doing:** one or the other or both of our children throw up every day, often more than once. Their medications make them sick, and it makes them sad to have to take it. I too am sad each time I give it to them, though I don’t let them know it. They take medications every six hours around the clock 365 days a year ... and my husband and I are TIRED.



Jenna and Patrick Partington

Our kids are so thirsty that one of their favorite gifts is a new water bottle. They sleep with these bottles every night and wet the bed all night long, creating no less than four loads of urine-soaked laundry every morning. No one wants to sleep on a wet bed, so we have medical-grade absorbent mats that we change several times each night. We are TIRED. I’m afraid of

colds, the flu, viruses and any illness that can affect our high-risk kids. It was four years ago, but I still remember our daughter in the intensive care unit – fighting for her life.

And while we are on hospitals: I am excited about the future but afraid of research studies and having young children who may be among the first to try new, unproven treatments. It’s all in the name of progress, but I am fully aware that nothing is risk free.

Jenna is so slight and fragile; mentally tough but physically weak. Patrick eats constantly – cystinosis is a metabolic disease and his metabolism operates at three times the rate of other kids so keeping him filled with the salt, water and calories he craves (and needs!) is demanding to say the least.

**How do you tell a six-year-old they have a terminal disease?**

**How do you explain that they will wet the bed until they are teens, perhaps beyond?**

**Will they join their school friends on overnight retreats or be too embarrassed to go?**

**Will we find a cure in time for them?**

Muscle weakness, muscle wasting, high cholesterol levels, rickets, kidney transplants, photosensitivity, dehydration, loss of sight, thyroid problems ...

# A Mother's Elevator Speech by Teresa Partington

these and other anomalies have already or will one day strike Jenna and Patrick.

**You ask: "How are you doing? How are the kids doing?" We are tired and we are scared. And we have so much HOPE!**

## Let's Talk About Dr. Stephanie Cherqui

First, may I add something that most summaries about Dr. Cherqui will not convey? She is darling! An absolutely charming mother of two, Stephanie Cherqui is an unassuming French doctor, who has made curing cystinosis a professional AND personal mission. She shares her discoveries in a thick French accent, bubbling over when she describes the mice she has cured of cystinosis.

### To summarize what we learned from Dr. Cherqui that weekend:

We may look forward to two types of Human Clinical Trial Stem Cell Transplants (i.e.: attempts to CURE cystinosis) in the coming years:

**Allogeneic** (a sibling donor stem cell) transplant would come first, and if successful would make possible the first **Autologous** (the own patient's stem cell) transplant.

If Stephanie's work continues to be successful, bone marrow stem cell transplants could one day be the preferred method of treating end-stage renal failure – in other words, stem cell transplants could replace kidney transplants.

The methods proposed for Stephanie's trials have already been

tested and FDA-approved and used in pediatric trials for other diseases. This saves years of research time and money, and carries less risk for the patients in human clinical trials.

**The first allogeneic stem cell transplant will likely take place within the year. May I repeat? Within this year!**

I would like to thank Dr. Cherqui for her dedication and brilliance. She takes care to make no promises; but she has knowledge, competence and determination that I truly believe in.

## The Cystinosis Research Foundation

Thanks to the Cystinosis Research Foundation and the families, friends, doctors and researchers who have supported it since 2003 the following has happened to date.

- CRF has funded 78 cystinosis studies in eight countries around the world.
- Nearly \$15 million has been raised by CRF since 2003. 100 percent of these funds have been used or committed to fund research.
- *Jenna & Patrick's Foundation of Hope*, inspired by our children in 2004, has contributed nearly \$1.1 million to CRF efforts. We choose to pass every dollar earned to CRF, which does an amazing job of doling out funds thoughtfully and discriminately under the guidance of its world-class Scientific Review Board.

## To the Parents Who Travel This Journey with Us

We need to work together to find a cure in time for all of our children. If you have a newly diagnosed child, or if you are feeling isolated as you deal with this rare, mysterious disease please know that there are families who want to help. **You can e-mail me at [tjpartington@sbcglobal.net](mailto:tjpartington@sbcglobal.net) with any questions.**

**As a parent you can be a big part of the cure by registering your child at [www.cystinosisregistry.org](http://www.cystinosisregistry.org).** Registering will help accelerate research, allow you to receive clinical trial announcements, learn about the cystinosis community and receive CCIR newsletters.

## Beyond Cystinosis

Medicine and the human body are such that the research done for one disease often leads to treatments for other diseases. I am proud that CRF-funded research has contributed to the study of Huntington's disease and NASH (fatty liver disease).

I am also proud that Stephanie's work may change the treatment of end-stage renal failure; perhaps one day bone marrow stem cell transplants will replace kidney transplants!

**I am overwhelmed by the ways (far more of them happy than sad) in which cystinosis has touched my life. Anyone reading this is part of a small, motivated group of people who set a five-year goal of curing cystinosis.**

*Hang in there kiddos!*

# Joshua Clarke

By Rose Clarke, Joshua's mom

It has been over 2 ½ years since Joshua was diagnosed with cystinosis. Much has transpired since that fateful doctor's appointment back in January 2008.

Joshua is now four years old and doing well. His year has been filled with many milestones. He recently "graduated" from physical therapy and has begun Tae Kwon Do classes. He has continued to gain weight and finally reached the growth chart for his height! Joshua has also made great strides

occupational therapy a week. He has made great progress with his eating difficulties; he continually asks for food and now is willing to try new foods and textures. He is, however, still unable to eat enough to sustain him. He relies on his g-tube for his daily nutritional needs and medications.



Our cookbook sold out in May, 2010. We sold 2,136 copies, which enabled us to contribute more than \$20,000 to CRF.

loves his Tae Kwon Do classes. He has been blessed with two siblings who want to play with him and always spoil him when they get the chance. Joshua has become a very accomplished helper around the house, enjoys music and has discovered the joy of video games.

Two and a half years ago our lives changed forever. In the months that followed his diagnosis, we were without hope and knew only despair. We have certainly come a long way.

With the help of the Cystinosis Research Foundation and all those involved with it, especially Joshua himself, we have much to be hopeful for. We are not immune to worries about the future with hourly eye drops to dissolve painful crystals, a renal transplant and a host of related organ malfunctions. We work each day and pray that the lifesaving medication, Cystagon®, will continue to prolong his life and prevent the horrible effects of this disease from causing Joshua any further damage.

We are very thankful for all those involved with the Cystinosis Research Foundation and the great strides they have already made for these patients. We are confident that a better quality of life is on the horizon for all cystinosis patients and that a cure is not far away.

**Thank you for your continual support of our family and Joshua's Journey of Hope.**



with his occupational therapy and will be starting pre-school in January.

Joshua currently takes eight different medications a day, every six hours. In addition, he is injected nightly with growth hormone. He continues to receive two hours of

Joshua remains a loving, energetic and joy-filled little boy. His beautiful spirit keeps our family smiling and laughing. Joshua enjoys sword fighting with whoever will spar with him, hopping in the car and going wherever and he absolutely

# Amazing Tina Flerchinger

By Denice Flerchinger

**L**ooking back ... 2010 was a year of change, with many challenges and blessings along the way!

Tina lost two grandfathers. There are no words to describe the adoration she had for them, and the love that was reciprocated was beautiful to witness. She enlightened me one day when she said, “Mom, don’t cry, we have two more angels looking over us.” Indeed, we must have with all that the year brought.

Tina and I spent most of September, October and November at Stanford Medical in Palo Alto, California. After five years of hoping and praying that Tina would one day be on this new “miracle” medicine (RP103) it finally came about. Tina was accepted into the FDA trial, weighing only one ounce more than the requirement. It was no easy task with the ongoing requirements, extended travel, being away from

home, missing school, and all the tests and blood draws Tina had to endure each day. But she did it so bravely – she continues to amaze us each and every day!

The RP103 has brought a dramatic improvements in Tina’s quality of life. The new medication is easier for her to tolerate – no more nausea and vomiting. She is off her stomach medications, has an appetite, and after six years is getting a full night’s rest.

In January, Tina lost her trademark when her gastronomy tube was removed. The “second belly button” has been a conversation piece for this social bug. No more

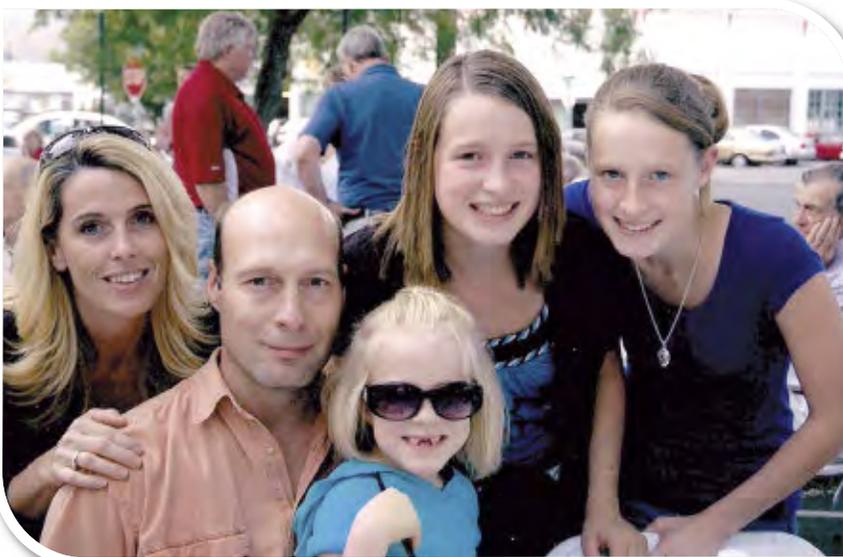


show-n-tells for this little sweetie.

I am so proud of Tina for all that she’s accomplished this year. She maintains a big toothless grin most days. She is such a joy to be around and completes our family perfectly. She is learning to read and loves it, along with first grade and all that it has to offer.



*It was no easy task with the ongoing requirements, extended travel, being away from home, missing school, and all the tests and blood draws that Tina had to endure each day. But she did it so bravely – she continues to amaze us each and every day!*

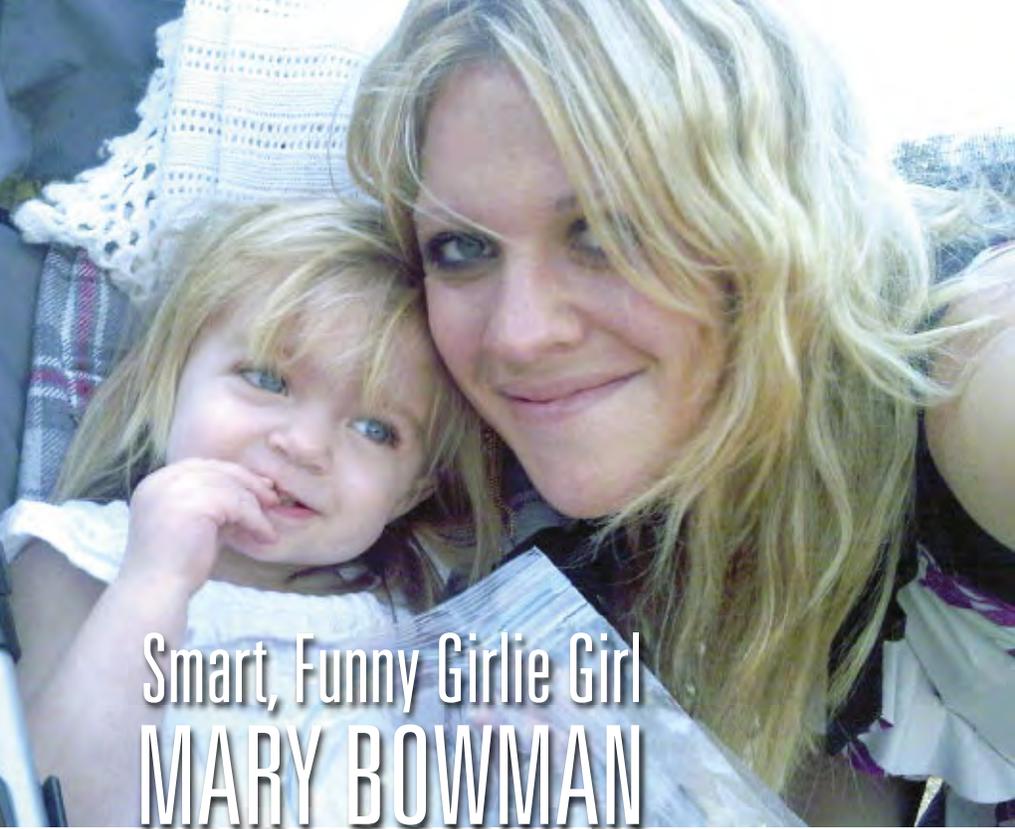


*Tina (center) with her mom Denice, dad Mark and sisters Catherine and Nichole Flerchinger.*

Tina still takes eight medications each day (18 pills and 16 doses of liquid meds) and has a multitude of other health issues, but we continue to count every blessing and miracle along the way!

A sincere “thank you” to Nancy and Zoe for being the crusaders they are, and all those who support the Cystinosis Research Foundation – donors, researchers, and our wonderful community of family and friends.

You have given us hope and we know one day Tina will be free from cystinosis.



# Smart, Funny Girlie Girl MARY BOWMAN

By Melissa Bowman-Head

Our youngest daughter Mary was born a full-term, seemingly healthy baby. She was actually the largest of our children! At about 7 months of age, Mary’s health began to decline. She started vomiting after almost every feeding, and vomiting bile in her sleep. She showed no interest in food and her growth and development stalled.

Alarmingly, one day we noticed numerous bulges in her skull. This was later diagnosed as rickets. After two months of testing and worry, Mary’s pediatrician was at a loss and told us if she didn’t gain a full pound in three days she would be admitted to Seattle Children’s Hospital, which is exactly where we ended up. After five days and numerous tests ruling out various diseases, her doctors were almost certain Mary had cystinosis. Her confirmed diagnoses was rickets, involving her skull and long bones, and renal Fanconi syndrome.

When Mary was sent home from the hospital, it was with an NG feeding tube and armed with formula, feeding pump, feeding bags, syringes and

medications. We faced a new, terrifying way of life. Three weeks later the diagnosis of cystinosis was confirmed. It was a relief to finally have an answer yet also so horribly frightening. Since Mary’s nutritional and medication requirements were now long term, we had her g-tube placed.

Mary continues to require frequent dosing of electrolytes throughout the day plus her Cystagon® every

six hours. She receives nutrients and calories 12 hours a night while she sleeps using a feeding pump.

Mary recently started using cysteamine eyedrops. She never fusses when she receives them, even though they sting – and sometimes she even asks for more.

Caring for Mary presents many challenges and requires support from my husband and our whole family. For me, fatigue is difficult to manage.

I am up every 2–3 hours to change Mary’s diaper and give her the nightly Cystagon® dose. Due to Fanconi’s, Mary is always thirsty and drinks two large cups of water every night.

Our family of five lives in rural Washington State. Mary loves her older sister Audrey (age 8) and brother Wylie (age 5). Her energy level and happiness skyrocketed when she began her supplements. She has developed into a smart, feisty, funny 2-year-old, always making us laugh, which is very different from our sickly baby. She loves Strawberry Shortcake, Dora the Explorer and playing with her baby dolls. She tries to put “eyedrops” in her baby’s eyes to make them “all better”!

She is quite the girlie girl already having preferences in her daily wardrobe and often brings me nail polish for her fingers and toes. Mary rarely eats, but she loves mealtimes and wants everything on her plate like her brother and sister. Her favorite foods are ketchup, peanut butter and ice cream!

Contending with cystinosis, it still a challenge, but it is more manageable. Since being diagnosed, we have spent almost two months in the hospital with complications from electrolyte imbalance and g-tube management.

➤ *We want a full, long life for Mary filled with joy and happiness just as we do for our other children.*

The current calm is welcome. Lab draws, which were previously done weekly, are now done once a month.

We are optimistic about Mary’s future, knowing that her health can be sustained with the current regimen of medication and supplements, and that the dream of a cure is becoming a closer reality.

**We want a full, long life for Mary filled with joy and happiness just as we do for our other children.**

# Graceful Gabbie Strauss

By Gabrielle's mom, Jody Strauss

Over the last year, we have watched Gabrielle grow into a creative and caring 3-year-old girl who loves to play, have fun and laugh with her younger sister, Chloe.

Since starting preschool last September, Gabrielle has developed a passion for painting. We can't seem to find enough places and spaces for all her artwork. Most recently, Gabrielle and Chloe have been enjoying outdoor activities together. They love to shovel snow (they each have their own red shovel), toboggan and ice skate.

Gabrielle has grown taller and stronger, and she was discharged from physiotherapy because of her increased strength and capability. She continues to eat well and take all of her medications orally without any problems. She faces the challenges of cystinosis with grace and bravery. I'm sometimes surprised that she isn't grumpy or annoyed that we wake her up in the middle of the night for meds. Instead, she smiles, sometimes waves goodbye or asks us to say prayers again.



Photo use courtesy of Waterloo Region Record

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



We count our blessings everyday. We are thankful for the Cystinosis Research Foundation, associated doctors and researchers and our amazing family, friends and community.

The Cystinosis Awareness & Research Effort (CARE) has received wonderful support in 2010. The *Real Men Can Cook* Committee of Waterloo Region selected cystinosis as their charity for 2010 and 2011. Last April, 100 men from our community took over the kitchen and became chefs for an evening of competition, food and fun. The Ontario Tire Dealer Association also held their annual charity golf event and raised \$36,000 for cystinosis research, which was matched by a local philanthropist. We are so grateful for the wonderful support we have received.

We count our blessings everyday. We are thankful for the Cystinosis Research Foundation, associated doctors and researchers and our amazing family, friends and community.

We continue to take one day at a time, putting our faith and trust in God to give us strength for the difficult times, hope for the future, and peace and joy through it all.

## UPDATE ON EYE DROPS



In September 2010, the U.S. Food and Drug Administration (FDA) issued a complete response letter to Sigma-Tau Pharmaceuticals, Inc. for CYSTARAN™ (cysteamine hydrochloride ophthalmic solution), a therapy for the treatment of corneal cystine crystals in patients with cystinosis.

The final approval has been delayed to allow the third party manufacturer of CYSTARAN™ to meet additional standards of production requested by the FDA. While the FDA's questions about manufacturing are impacting the timeline for product availability, Sigma-Tau Pharmaceuticals, Inc. is committed to the cystinosis community and is diligently working to reach the final stage of the product's approval process.

**For more information regarding CYSTARAN™ and its pending FDA approval, please contact Lesli King at [lesli.king@sigmatau.com](mailto:lesli.king@sigmatau.com).**



We have so much to be thankful for this year. Bailey recently finished the RP-103 study at Stanford Hospital.

## Our Hero, Bailey DeDio

*By Jessica, Bailey's mom*

**W**e are so excited because Bailey was given the RP-103 medication on the last day of the study. He now takes RP-103 two times per day, instead of Cystagon®, which he took four times per day. I cannot tell you how much the new medication has changed our lives. Bailey can sleep through the night now, his stomach feels better and he takes one less medication.

Bailey is a new young man, he feels more confident and independent. He is in seventh grade and doing great. He recently got a Honda 85 dirt bike that he loves to ride. He is on a bowling league and loves to be around his family and friends. We are so proud of Bailey, he is our hero!



*We cannot express our gratitude, love and appreciation enough for the Stacks. Thank you Natalie for having a WISH! Thank you Raptor, doctors, nurses and all the families with cystinosis we were blessed to meet on Stanford journeys. We love you all!*



*Bailey, Jay, Jessica and Ryder DeDio.*



# Nicole Hall

## Growing Bolder, Stronger and More Hopeful Every Day

Nicole Hall turned five in October and started school last fall. We enrolled her in Pre-Kindergarten, where she is still the smallest child in her grade, but she is fitting in very well. Being in school has been wonderful for her and she really loves her class. She enjoys learning new things and then coming home and teaching our family.

School has also provided her a great opportunity to develop socially and she has made some very good friends. It is amazing how much she has come out of her shell over the past five months. She is still shy with strangers, but has started warming up to people quicker.

The school has been very supportive of Nicole's special needs, which includes feeding her through her g-tube and giving her eye drops to prevent crystals from forming in her eyes. The school also gives Nicole her mid-day dose of Cystagon®, which needs to be taken every six hours – and is one of the 14 doses of medication she receives every day. We are very excited about the FDA approval of the delayed-release cysteamine, which means we will only need to give Nicole medicine twice a day.

Nicole was diagnosed with cystinosis in August 2007, two months before her second birthday. She started showing symptoms when she was four months old, when she began

to fall off the growth chart. It took countless doctors visits, tests and hospital stays over the next 18 months before she was diagnosed with this rare disease.

Nicole has made a lot of friends at school, but her best friend is still her sister, Angie, who will turn four in April.

When Nicole gets home from school, you can find the two of them playing pretend – whether it is with their Barbies, Polly Pockets or My Little Ponies. They also love to color and draw pictures for mommy and daddy.

The strength Nicole shows each day makes it obvious that she isn't letting this disease run her life. She has her bad days, but she fights through them and always keeps a positive attitude.

We are grateful

for all of the work of the Cystinosis Research Foundation and to everyone who supports it. We are also thankful for the wonderful people who are working to find better treatments and eventually a cure.

*Aaron, Stephanie, Nicole and Angie Hall*



*On December 5, the Third Annual Running for Nicole fundraiser was held to raise money for CRE. This year, we had two teams in the Dallas White Rock Marathon relay. Each team beat their goal times making it a great success! The guy's team was comprised of Geoff Nenninger, Wade Clerkin, Jim Plutt, Michael Galiunas, and Aaron Hall. The women's team was run by Becca Smith, Alissa Nenninger, Suzanne Clerkin, Kristen Brom, and Stephanie Hall. We were blessed with donations of more than \$12,250. As Nicole grows, so does the event – in 2011 we will have three teams in the relay!*



# Henry and Friends

Participants at the starting line in the *24HoursforHank*  
*Cycling for Cystinosis* event in September 2010.



# Race Towards a Cure

2010 was a busy, fun-filled year for Henry, and his family and friends. And it was also a year filled with many blessings.

Henry is 4 ½ now and doing well. He is in preschool and attends occupational, physical and speech therapies twice a week. We're still working on potty-training, but hopefully that will happen soon.

Henry receives growth hormone shots six nights a week, which has helped him grow 3 ½ inches and gain strength. His self-confidence, balance and coordination have increased significantly, and he can now snow ski by himself with his poles. He is proud of his accomplishments and so are we!

Henry doesn't have much of an appetite, but we are excited about the new delayed-release cysteamine. Hopefully the new drug will allow him to eat more often, which should prevent him from feeling sick four times a day. Henry is practicing swallowing small pills so he can take the new medication when it's available.

2010 was a busy year for our foundation, *24HoursforHank*. In January, our 24-hour ski event, *24 Hours of Schweitzer*, which also featured a dinner and an auction, was a big success with over 100 participants. We also held our 24-hour cycling event, *Cycling for Cystinosis*, in September. 2010 was the first year we hosted both events, so a big thank you to our board of directors is in order! We are also thankful to all of our supporters, family, friends, participants and volunteers. They make everything we do possible, which gives us a chance to feel like we are making a difference by helping the Cystinosis Research Foundation in its quest for better treatments for cystinosis and ultimately a cure.



Skiers at the starting line at *24 Hours of Schweitzer*.



Our doctors and nurses have been wonderful too. Since Henry sees so many medical professionals, it is great having such good relationships with them all, and knowing they work together so well.

We would like to say a special thank you to the Cystinosis Research Foundation and its founders Jeff and Nancy Stack, for their dedication to making the lives of everyone who faces this disease immensely better. We are forever grateful for the hope they have given our family!

*Tricia and Brian Sturgis*

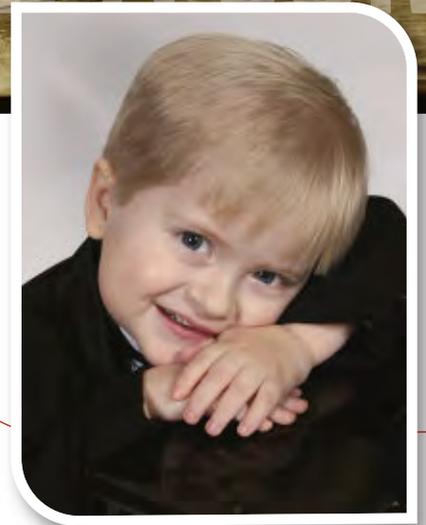
By John Sanders, Camden's dad

## HELP CAMDEN FIND A CURE

Camden was born on October 24, 2005. The first thing we noticed about Camden was that his hair was bleach blond, almost white. He seemed normal for the first few weeks – some spitting up and throwing up and sometimes not sleeping well at night, but we did not think much about it. I can remember nights when my wife, Amanda would wake me up in tears, exhausted from trying to satisfy him. He would drink a bottle and throw it up but then a second bottle would stay down. When we told the doctor about it, he blamed it on reflux. He also said that we were spoiling Camden, which is why he wasn't sleeping at night. Fortunately, we didn't listen to that doctor and continued to feed Camden and take

on a regular basis – Sour Cream & Onion and Salt and Vinegar are his favorites. He also liked hot sauce and spicy foods. We thought it was strange that he liked those foods but we knew they wouldn't hurt him.

Camden did not sit up or walk on time. He was almost a year old when he sat up and several months later he started walking. Again, we took him to the doctor because he continued to throw up so often and he urinated frequently. The doctor checked him for diabetes but his sugar levels were fine. If the doctor had drawn blood and done a full panel on him, he would have discovered that his electrolytes were out of whack and that might have helped with a diagnosis.



potassium level was so low he might have a heart attack.

For two days one specialist after another examined Camden. They all gasped in amazement at how well he was doing. Dr. Warshaw and Dr. Riar were great to us the whole time we were there.

We started Camden on Cystagon® and several other medications. The doctor warned us about the taste and the smell and told us he might eventually refuse it. Camden initially took the medications but in October, he refused to take them. We had to increase his dose and finally had to have a g-tube put in. The g-tube has been a blessing. Camden now eats regular meals. I give him instant potatoes and whatever else I can blend up and can put in the g-tube. Two weeks after the g-tube was in, Camden gained two pounds!

I used to be really rough on Camden and my wife was easy on him. Now it's the other way around. He is my little heart and I feel like he is the reason I am still here – to take care of him. I get up every night to give him water, milk and his meds. The doctors think that Camden might have a less severe form of cystinosis because he does nothing but drink water.

There would be nothing better than to find a cure for cystinosis. It is my goal to make sure Camden lives a long life even if it means he gets both of my kidneys. Children should not suffer.



*More than 350 people attended the Help Camden Find A Cure, which raised almost \$11,000!*

care of him. We put him on soy milk, which helped.

When Amanda went back to work, Camden was with two different babysitters. He liked one of them a lot and we now call her our “Angel from Heaven.” We would come home and the sitter would have Camden on the counter eating salt out of the shaker and butter by the spoonful. Come to find out, that was what he craved. He ate chips

Eventually, we changed doctors. The new doctor, Dr. Slade, examined Camden, listened to our reports and said, “I think he's fine but let's get a second opinion from a specialist in Atlanta.” In February, 2010, when he was 4<sup>1/2</sup> years old, Camden was finally diagnosed with cystinosis. My wife and I were shaken up beyond belief. We came to the hospital for a simple appointment but were told we had to stay because Camden's

When a baby is born, so many life changes take place. For me, I instantly believed there was something out there greater than myself.

Our son Skyler changed our life the moment he was born. My husband Van and I became a family. Being only 20 years of age, we instantly grew up and we realized that Skyler is a miracle that has been gifted to us for a reason.

However, we knew something wasn't right even on the night we brought him home. Skyler cried all day and night. At four months he showed his first symptoms of cystinosis. He was hungry and thirsty all the time, but he wasn't digesting his food or gaining weight because of frequent vomiting. He was miserable. There were times when I could do nothing for him, and I would hold him tight and just cry with him and tell him we'd have to get through this together.

After seeing many doctors, in desperation we took Skyler to the hospital and the nightmare began. At one year old he weighed only 11 pounds. He had developed four staph infections. He became septic and we almost lost him. I remember kneeling in the ICU waiting room begging God not to take him. Amazingly, my prayers were answered and Skyler recovered from his infections. A second miracle!

He stabilized and we brought him home but we still had no diagnosis. We knew he was in kidney failure and had gastric issues, but nothing else. He had tubes coming out of everywhere but we were home! Home! It was the best feeling in the world to us.

We spent our days cleaning up vomit, tube feeding and trying to get Skyler to eat. We endured many doctor visits, surgeries and blood



By Christina Minella, Skyler's mom

## A Miracle Named Skyler

*Skyler Minella with his dad Van and his mom Christina.*

draws. Understandably, we celebrated every milestone.

Desperate to find a diagnosis, my husband searched Skyler's symptoms on the internet and found cystinosis. We showed it to Skyler's kidney doctor and at 18 months old he was diagnosed with cystinosis. We were so thrilled to have a diagnosis and knew it was time to get educated about the disease so we could do everything in our power to help him.

the stomach but we stuck with it.

Skyler is now 7 years old. His kidney condition has reversed itself, leaving his doctors stumped. His kidney function is an incredible 100 percent! Can you believe it? It reversed itself! A third miracle has happened!

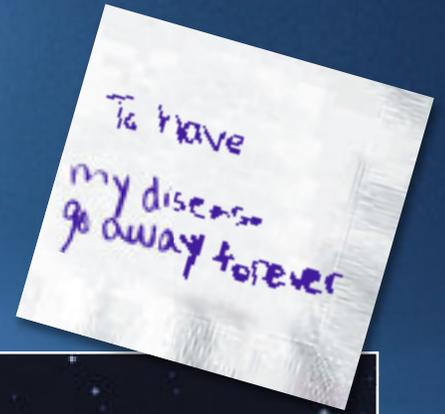
Skyler is our only baby and with three miracles we feel so blessed to have him in our lives. Our lives are so full of blessings that I became a photographer to capture all the great-

➤➤ *I remember kneeling in the ICU waiting room begging God not to take him.*

Doctors and the cystinosis support group told us about Cystagon® and how important it was to keep his levels correct and give the medication on time. Cystagon® is taken every six hours, which meant Skyler would never get a full night's sleep. I remember how blessed we felt that he was alive so this seemed like a piece of cake to us. Side effects are extremely hard on

est moments of his life. His infectious laugh puts you in a good mood no matter what kind of day you're having. When he's feeling good he has an amazingly vibrant personality and the eyes and smile of angel. Our only fear now is that cystinosis will take his life at an early age, so we now pray for a fourth miracle. We pray for a cure!

On the eve of her twelfth birthday, Natalie Stack  
made a wish no child should ever have to make.  
*"to have my disease go away forever."*



SAVE THE DATE  
ANNUAL NATALIE'S WISH EVENT  
SATURDAY, MAY 21, 2011

*The Wish Heard  
Round the World.*

**WE INVITE YOU TO JOIN US AT THE BALBOA BAY CLUB FOR  
OUR 2011 NATALIE'S WISH EVENT ON SATURDAY, MAY 21.**



OVERTONE

The event will feature international singing sensation OVERTONE who recently performed at the Wynn Resort in Las Vegas and who was featured in the Clint Eastwood film *Invictus*.

That simple but heartbreaking wish – scribbled on the back of a restaurant napkin – has understandably become the wish of every child and adult with cystinosis. Today, Natalie's wish has also become a powerful rallying cry for friends and supporters of the Cystinosis Research Foundation from around the world.

Given the extraordinary successes that have already been achieved through CRF efforts, this year's celebration will undoubtedly be the most exciting and emotional ever.

For sponsorship information or reservations contact Zoe Solsby at [zsolsby@cystinosisresearch.org](mailto:zsolsby@cystinosisresearch.org) or 949-223-7610.



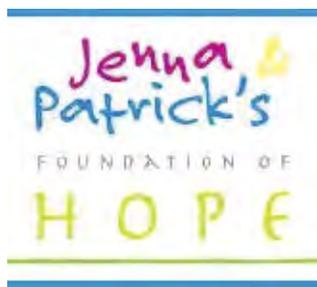
NEVER DOUBT  
that a small group of  
thoughtful,  
committed citizens  
can change  
the world.  
INDEED  
it is the only thing  
THAT EVER HAS.

*Margaret Mead*

Some families and friends run for a cure while others sell cookbooks, hold bake sales, golf tournaments, dinners or ski-a-thons. The events take place in rural communities and in large metropolitan centers. Some involve small numbers of people while others involve hundreds and maybe even thousands.

It all just proves that there are many ways to raise money for a wish – *to have my disease go away forever* – that has touched so many throughout the world.

Here are some of CRF's partners who have contributed to our success over the years. Your generosity through their remarkable efforts has helped us find better treatments and move closer than ever to finding a cure for cystinosis.



**Jenna & Patrick  
Partington  
\$1,079,301**



**Holt Grier  
\$485,170**



**Henry Sturgis  
\$200,050**



**Gabbie Strauss  
\$85,988**



**Tina Flerchinger  
\$78,038**



**Joshua Clarke  
\$39,255**

**\*The Cystinosis Foundation – \$20,000**

**Oliver Britten – Letter Writing Campaign – \$35,015**

**Nicole Hall – Running for Nicole – \$32,492**

**Camden Sanders – Help Camden Find a Cure – \$10,750**

**Katie Ahnen – North Face 50 Mile Trail Run – \$4,628**

*We genuinely appreciate your continuing support.*



THIRD INTERNATIONAL  
Cystinosis Research Symposium

LEARN ★ SHARE ★ COLLABORATE

**The Cystinosis Research Foundation is proud to announce that its third International Research Symposium will be held on Thursday, March 29 and Friday, March 30, 2012.**

The CRF is committed to advancing cystinosis research. The 2012 CRF symposium will allow clinicians, scientists and doctors from around the world the opportunity to meet and discuss current research directed toward improved treatment and a cure for cystinosis.

**THE SYMPOSIUM WILL:**

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

**BECKMAN CENTER OF THE NATIONAL ACADEMIES OF ENGINEERING AND SCIENCE**

Irvine, California 92617  
[www.nationalacademies.org/beckman/](http://www.nationalacademies.org/beckman/)

More information will be available later this year. Contact Foundation Chair **Nancy Stack** at **949-223-7610** or **nstack@cystinosisresearch.org**.



**CYSTINOSIS**  
RESEARCH FOUNDATION

## International Symposium Brings Out the Best in Cystinosis Researchers *By Dennis Arp*

A symposium that illuminated the latest medical advances also honored a guiding light who has dedicated his career to improving the lives of those with cystinosis.

Dr. Jerry Schneider’s unparalleled impact will long inspire the research and clinical communities, just as his insights imbued the Second CRF International Cystinosis Research Symposium, for which he served as chairman.

The two-day event featured tributes by everyone from Cystinosis Research Foundation (CRF) Chair Nancy Stack to patients and family members to medical colleagues of Dr. Schneider, who has retired after 45 years as a tireless advocate for cystinosis research.

The fruits of Dr. Schneider’s influence were reflected in the work presented at the symposium, held April 8–9, 2010, at the Arnold and Mabel Beckman Center of the National Academies of Sciences and Engineering in Irvine.

More than 60 scientists and researchers from eight nations attended the event, which was underwritten and organized by the CRF. Participants presented their research progress and exchanged ideas, including those that hold the most potential to cure this rare and fatal disease.



*It was unmistakable that the symposium fostered a dynamic synergy among the researchers.*

All of their research work is supported by CRF grant funding.

“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. “Other investigators horde, cystinosis researchers share.”

Gene therapy is one of the most promising areas of research, and symposium attendees learned that a group of leading researchers has formed a consortium to advance this field of study. “We are dedicated to bringing to reality the first stem cell and gene therapy clinical trial for cystinosis,” said Stack, co-founder of the CRF with her husband, Jeff. Nancy also chairs the CRF Board of Trustees.

Also attending were officials from sponsors Sigma Tau Pharmaceuticals and Raptor Pharmaceuticals, which is conducting Phase 3 trials on delayed-release DR Cysteamine, a breakthrough medication developed thanks to CRF grants.

“All the researchers who attended are passionate about their research, committed to our children and dedicated to understanding more about cystinosis,” Stack said. “The scientists shared their data and discoveries, debated new theories and formed collaborations. It was unmistakable that the symposium fostered a dynamic synergy among the researchers.”

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**To ensure continued research progress, the foundation has announced that the third CRF International Cystinosis Research Symposium will be held March 29–30, 2012.** *(See sidebar at left for details)*

# Research Progress Fuels Real Hope for a Cure

By Dennis Arp

**Energized by achievements that buoy the entire cystinosis community, researchers harbor a resolve to find a cure and better treatments that has never been stronger.**

**And the best news is that the future looks even brighter.**

“We are moving closer every day to a cure for cystinosis,” said Nancy Stack, co-founder of the Cystinosis Research Foundation and chair of its Board of Trustees. “We are extremely fortunate to fund the most talented scientists in the world who are dedicated to saving our children.”

**At The Scripps Research Institute in San Diego, scientists have made extraordinary discoveries using gene and bone marrow therapy that may forge a pathway to a cure.**

“In other human disorders, a person’s own stem cells have already been used safely,” said Dr. Stephanie Cherqui, an assistant professor at Scripps who is leading the project. “For cystinosis patients, this strategy might create a reservoir of healthy stem cells in the bone marrow during the patient’s lifetime. Hopefully these cells will respond to tissue damage and travel to repair the different organs of the cystinosis patient.”

Dr. Cherqui and her colleagues have already enjoyed success reversing cystinosis in laboratory mice, and the new CRF Cystinosis Gene Therapy Consortium is working to translate these results into an FDA-approved clinical trial. “If all goes well, there will be a clinical trial for a cure within the two to four years,” said Stack, a consortium member.

Closer on the horizon Raptor Pharmaceuticals is in Phase 3 and Phase 4 clinical trials with DR Cysteamine, which reduces patient dosing from every six hours to every 12 hours. The delayed-release medication means cystinosis patients can experience the joy of actually sleeping through the night.

## CRF Science Report and Research Grant Updates

CRF research grant progress reports are published in the *Research* section on our website: [www.cystinosisresearch.org](http://www.cystinosisresearch.org). As updates are received they appear in our monthly *Star Facts* e-newsletter. To receive these updates or a printed copy of our *Science Report*, please email your contact information to [zsolsby@cystinosisresearch.org](mailto:zsolsby@cystinosisresearch.org).

## 2011 Call for Funding Proposals

The ultimate goal of the Cystinosis Research Foundation is to find a cure for cystinosis. Global calls for grant applications will be 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 three years.

Currently, the CRF has more than \$1.5 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](http://www.cystinosisresearch.org) for details.**

## 2010 Research Grants Funded

**Total: \$1,014,970**

**Alan Davidson, PhD, Principal Investigator**  
University of Auckland, New Zealand  
“Characterization and Rescue of CTNS-iPS Cells”  
\$129,557 – 1-year grant

**Bruno Gasnier, PhD, Principal Investigator**  
Institut de Biologie Physico-Chimique,  
Paris, France

**Ellen Closs, PhD, Co-Investigator**  
University Medical Center, Mainz, Germany  
“Molecular Study of Lysosomal Transporters Involved in the Cystine-depleting Effect of Cysteamine”  
\$169,384 – 2-year grant

**Patrick Harrison, PhD, Mentor**  
**Ciaran Lee, Research Fellow**  
University College Cork, Ireland  
“Cystinosis Gene Repair”  
\$146,258 – 2-year grant

**Elena Levchenko, MD, PhD, Principal Investigator**  
University Hospital Leuven, Belgium  
“Studying Podocyte Function in Nephropathic Cystinosis”  
\$150,000 – 2-year grant

**David Pearce, PhD, Principal Investigator**  
University of Rochester Medical Center,  
Rochester, New York  
“Yeast Model for Cystinosis”  
\$26,211 – 6-month grant

**Jennifer Simpson, MD, Principal Investigator**

**James Jester, PhD, Co-Investigator**  
University of California, Irvine  
“Novel Treatment Modalities for Corneal Cystinosis”  
\$180,000 – 2-year grant

**Mary Taub, PhD, Principal Investigator**  
State University of New York at Buffalo  
“Mechanisms Underlying the Fanconi Syndrome in Cystinosis”  
\$104,067 – 1-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

### CHAIR

**Nancy Stack**  
Cystinosis Research Foundation

### VICE CHAIR

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

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**Tricia Sturgis**

24 Hours for Hank

**Zoe R. Solsby**

Cystinosis Research Foundation

## MEDICAL AND SCIENTIFIC ADVISORY BOARD

**Stephanie Cherqui, PhD**

The Scripps Research Institute  
La Jolla, California

**Ranjan Dohil, MD**

University of California, San Diego  
San Diego, California

**Allison A. Eddy, MD**

Seattle Children's Hospital  
Seattle, Washington

**Robert Mak, MD**

University of California, San Diego  
San Diego, California

**Jerry Schneider, MD**

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

**Jennifer Simpson, MD**

University of California, Irvine  
Irvine, California

**Doris Trauner, MD**

University of California, San Diego  
San Diego, California

## 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

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  
Rome, Italy

**Julie R. Ingelfinger, MD**

Harvard Medical School  
Boston, Massachusetts

**Elena Levtchenko, MD, PhD**

University Hospital Leuven  
Leuven, Belgium

**William Rizzo, MD**

University of Nebraska Medical Center  
Omaha, Nebraska



**CYSTINOSIS**  
RESEARCH FOUNDATION

# Denice Flerchinger and Whitney Glaize

## Two Very Special Women Join the CRF Board of Trustees



I am Denice Flerchinger, and in February 2005 my life changed with one word: “cystinosis.” My youngest daughter, Tina, was diagnosed with cystinosis at the age of 17 months. After processing this information, and settling into our new lifestyle, my family and I started meeting with other children and adults with cystinosis.

It soon became apparent that many parents struggle to adjust to the needs of caring for a child with cystinosis. The diagnosis comes with so many challenges – preparation and scheduling of medications, dealing with physicians, pharmacies and insurance companies, special school needs – not to mention the often unacknowledged emotional needs of the caretaker.

**I made a pact to do everything I could to help these families. This has become my focus and my passion. If I can help just one family I'll feel I've done my job.**

Therefore, it was a great honor when I became a trustee of the Cystinosis Research Foundation in 2010. I look forward to serving the cystinosis community in a multitude of ways. I serve on the board of *Tina's Hope for a Cure*, a nonprofit foundation started in honor of my daughter to raise money for cystinosis research. Our foundation has done an outstanding job of raising awareness in our small community of 40,000, as well as raising monies to help fund a cure.

My husband, Mark, and I own an electrical contracting business. I am vice president and chief financial officer. I integrate accounting and business management, and work with budgets, develop business strategies and manage employee relations.

Every day brings a new challenge and I love what I do, but my most important “job” is being a mom to our three daughters. Nichole and Catherine attend Clarkston High School, and Tina attends first grade at Cornerstone Christian School.

In addition to volunteering for various organizations, I enjoy cooking, traveling, reading and spending time with our large families – with a total of 11 siblings, 38 nieces and nephews, and 7 Godchildren.

I am motivated, reliable, flexible and detail-oriented. I have integrity in my work, honesty in my words, and feel like I will bring a keen insight to the CRF. I recognize the importance of uniting the cystinosis community, through the registry and other means, so we can all work more effectively toward our goal of finding a cure. **I would love to play a part in putting an end to this terrible disease.**



Whitney Glaize is a Speech Language Pathologist currently working in Orlando, Florida.

She primarily works with the pediatric population. She specializes in preschool language disorders, augmentative and alternative communication, autism, and feeding and swallowing disorders.

She received her master's degree from University of Central Florida in 2003 and her Bachelor's degree from University of South Florida in 2001. She has worked in the field of speech therapy since 2001.

Whitney was diagnosed with cystinosis in 1980 and has been on some form of cysteamine for the last 30 years and is one of the only adults who has yet to receive a kidney transplant. She began supporting the Cystinosis Research Foundation in 2006 and is passionate about finding better treatment options. Her dream is to one day be cured of cystinosis.

**Living with cystinosis, together with her knowledge of the maladies associated with the disease, and the needs of both children and adult patients, make Whitney a perfect CRF board member, as well as a spokesperson for the foundation.**

She is ready and willing to advocate for children and adults fighting cystinosis and she is excited about the amazing research that the CRF has funded through the years.



# 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, Innolyt, 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](http://www.cystinosisregistry.org)

## Allison A. Eddy, MD Joins CRF Scientific Review Board

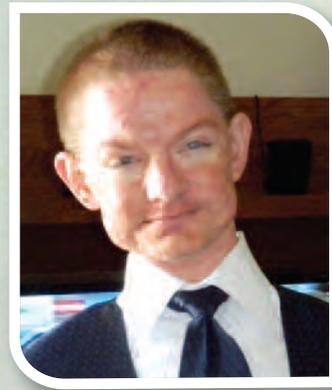
Allison A. Eddy, MD is a graduate of McMaster University Medical School in Canada. After completing a Pediatric Residency at Montreal Children's Hospital, McGill University and a fellowship in Pediatric Nephrology at the University of Minnesota in Minneapolis, she became a member of the faculty at the University of Toronto and a nephrologist at the Hospital for Sick Children for 12 years.



Allison A. Eddy, MD  
Professor of Pediatrics,  
University of Washington

In 1997, she was recruited to Seattle as Professor of Pediatrics at the University of Washington and Head of the Division of Pediatric Nephrology at Children's Hospital and Regional Medical Center in Seattle. In addition to clinical and administrative responsibilities, Dr. Eddy has had a long-standing interest in training nephrology fellows and in basic science research. She has directed pediatric nephrology fellowship programs for 20 years, overseeing the training of more than 60 fellows. In Seattle she is the principal investigator of a NIH-funded research-training grant in pediatric nephrology and the program director of a NIH-funded Child Health Research Center. Dr. Eddy has been an independent investigator for 25 years and is internationally recognized for her work on the cellular and molecular basis of kidney fibrosis and progressive kidney disease.

She currently directs a group of 10 people in the pediatric nephrology basic science research. She was appointed as the first Director of the Tissue and Cell Biology Research Center at Seattle Children's Research Institute in 2007. From 2001–2007 she was the Deputy Editor of the *Journal of the American Society of Nephrology*. In 2009, she was elected to the Council of the International Society of Nephrology.



## Good-bye to My Dear Son

By Barb Brauer, Preston's mom

My 25-year-old son, **Preston Towriss** had all the normal symptoms of cystinosis: Fanconi syndrome, failure to thrive and small in stature. He took growth hormones and had a kidney transplant at age 7. He was an excellent patient who rarely complained and always had a smile.

He completed two years of college, wanting to be a mortician. He had his own car and apartment, and he was an avid sports collector who had memorabilia from almost every sport. Many of the items were personally autographed to him. He had an entire room dedicated to Shaquille O'Neal. Preston liked Shaq because of his height and because he helped kids. He loved his family, his friends and his cat, Hank.

The atrophy in his body, starting with blindness, forced Preston to quit driving and work around the age of 22. Difficulty with swallowing and weakness forced him into a wheel chair.

Preston was an inspiration to many. His love of God and his faith are what got him through to the end. He knew that although not perfect he was capable of living for the Lord. He prayed for his friends and prayed often for a cure for cystinosis so kids wouldn't suffer.

I believe his message would be: Be happy, love God and you will be perfect in heaven. He passed away August 22, 2010. He is now perfect.

# CYSTINOSIS FAMILIES UNITED FOR THE CURE

# Save the Dates

**Friday, April 1 – 2, 2011 ★ 8:30 am**

24 Hours for Hank – Henry Sturgis  
*The 3rd Annual 24 Hours of Schweitzer*  
Schweitzer Mountain Resort, Sandpoint, Idaho  
Information: [www.24hoursforhank.org](http://www.24hoursforhank.org)

**Saturday, April 2, 2011 ★ 6 pm**

Cystinosis Awareness & Research Effort – Gabbie Strauss  
*Chase the Cure @ Real Men Can Cook*  
St. George Hall – Waterloo, Canada  
Information: [Jody.Strauss@yahoo.com](mailto:Jody.Strauss@yahoo.com)  
[www.cystinosis.ca](http://www.cystinosis.ca)

**Saturday, April 16, 2011 ★ 7 pm**

Addison's Angels – Addison Cox  
*Toast for a Cure*  
Wine 101, Wake Forest, North Carolina  
Information: [www.addisonsangels.org](http://www.addisonsangels.org)

**Thursday, May 19 – Saturday, May 21, 2011**

Cystinosis Research Foundation  
*Day of Hope Family Conference*  
Balboa Bay Club, Newport Beach, California  
Information: [nstack@cystinosisresearch.org](mailto:nstack@cystinosisresearch.org)  
[www.natalieswish.org](http://www.natalieswish.org)

**Saturday, May 21, 2011 ★ 6 pm**

Cystinosis Research Foundation  
*2011 Natalie's Wish Event*  
Balboa Bay Club, Newport Beach, California  
Information: [Zoe.Solsby@cystinosisresearch.org](mailto:Zoe.Solsby@cystinosisresearch.org)  
[www.natalieswish.org](http://www.natalieswish.org)

**Saturday, June 11, 2011 ★ 6 - 9 pm**

Tina's Hope for a Cure – Tina Flerchinger  
*3rd Annual Wine, Stein & Dine*  
Rogers Toyota Showroom, Lewiston, Idaho  
Information: [Denice.Flerchinger@gmail.com](mailto:Denice.Flerchinger@gmail.com)  
[www.tinashopeforacure.org](http://www.tinashopeforacure.org)

**Wednesday, July 27 – Tuesday, August 2, 2011**

Tina's Hope for a Cure – Tina Flerchinger  
*Golf for the Guinness Book of World Records*  
Lewiston Golf and Country Club  
Information: [Geno.Bonnalie@hotmail.com](mailto:Geno.Bonnalie@hotmail.com)

**Wednesday, August 17, 2011 ★ 8 am**

Proceeds to Cystinosis Awareness & Research Effort – Gabbie Strauss  
*Dundee Wealth Management Charity Golf Event*  
Listowel, Ontario, Canada  
Information: [Jody.Strauss@yahoo.com](mailto:Jody.Strauss@yahoo.com)

**Saturday, October 8, 2011 ★ 8 am**

Addison's Angels – Addison Cox  
*5K Run/Walk*  
Downtown Wake Forest, North Carolina  
Information: [Nicole.Cox@addisonsangels.org](mailto:Nicole.Cox@addisonsangels.org)  
or 919-606-0600  
[www.addisonsangels.org](http://www.addisonsangels.org)

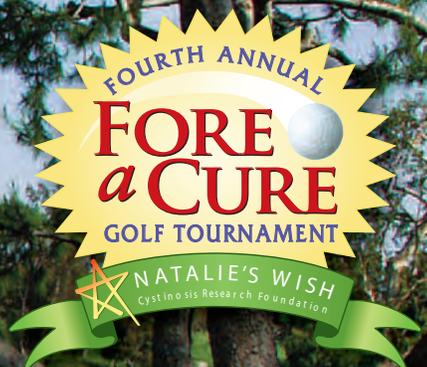
**Ongoing Fundraiser**

Skyler Minella  
*Hope for a Better Sky*  
Visit: <http://hopeforabetersky.blogspot.com/>

Connect with us at [www.cystinosisresearch.org](http://www.cystinosisresearch.org) or at  [www.facebook.com/CystinosisResearchFoundation](http://www.facebook.com/CystinosisResearchFoundation) 

## Save the Date: Monday, November 14, 2011

The Fourth Annual **Fore-A-Cure Natalie's Wish Golf Tournament** will be held Monday, November 14 at the exclusive Santa Ana Country Club. Plan to join us and many top Orange County business and community leaders for a perfect day of golf that you won't want to miss.



For sponsorship information or reservations, email [zsolsby@cystinosisresearch.org](mailto:zsolsby@cystinosisresearch.org)