



When your child's future is uncertain, hope, community and prayer sustain you.

# CYSTINOSIS RESEARCH FOUNDATION

We have come so far in the last few years and are moving closer to our mission, to find better treatments and a cure for cystinosis.

*We have much to be thankful for...*

- Our annual Natalie's Wish event in April raised \$1.6 million thanks to the 320 generous family and friends in attendance and the generous supporters who were unable to join us. The evening was highlighted by an amazing performance by **John Ondrasik of Five For Fighting**, who also contributed to the live auction with a stellar rendition of *Rocket Man* promoting the Elton John package that sold for \$10,000!
- **Jenna & Patrick's Foundation of Hope** held its Second Annual Swing & Bling Event on Friday, October 8, 2010 at the Citizen Hotel in Sacramento. The event honored Kevin and Teresa and their 5-year-old twins, Jenna and Patrick, who were diagnosed with cystinosis in 2006. The day of golf and evening festivities were sold out and raised more than \$250,000 for cystinosis research.
- In September, the CRF announced that \$750,000 was available for **cystinosis research proposals and fellowships**. After evaluation and review by our Scientific Review Board, we will announce the approved grants by the end of the year.
- Raptor Pharmaceuticals is completing the **DR Cysteamine Phase 3 Clinical Trials** at three locations in the United States: **Emory University**, Atlanta, Georgia; **Children's Memorial Hospital**, Chicago, Illinois; and **Stanford University**, Stanford, California. The **European locations** were just announced and include four sites in France and one in the Netherlands. We are proud to report that as a result of your generosity and commitment, the Cystinosis Research Foundation provided funding for every bench and clinical research study leading to the discovery of the delayed-release medication. We are optimistic that the FDA will approve DR Cysteamine in 2011.
- **CRF Cystinosis Gene Therapy Consortium** – the CRF along with a group of leading researchers and scientists have formed the CRF Cystinosis Gene Therapy Consortium. The goal of the 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 CRF is dedicated to bringing the first stem cell and gene therapy clinical trial for cystinosis to reality in the next three to four years.
- The CRF now has a Facebook page. Please visit us at [www.Facebook.com/CystinosisResearchFoundation](http://www.Facebook.com/CystinosisResearchFoundation) Facebook allows us to better connect with families and partners and quickly share the latest news about cystinosis. Join our group, "like our page" and follow us to stay up to date! (The links will always be on the CRF homepage).
- To better communicate with our cystinosis community, we have created the *CRF Star Facts* news brief. This e-newsletter is sent monthly with research updates and highlights events around the country to help fund cystinosis research. **To be added to the distribution list, contact us at [info@cystinosisresearch.org](mailto:info@cystinosisresearch.org)**

## Cure Cystinosis International Registry

We are pleased to announce the launch of the Cure Cystinosis International Registry (CCIR). The Cystinosis Research Foundation in partnership and collaboration with the Cystinosis Foundation and twelve other cystinosis family foundations from around the world, have

established a comprehensive, global patient registry for cystinosis. The CCIR questionnaire was developed by medical and scientific experts specifically for the cystinosis community. It is our mission to register every person with

cystinosis, thus connecting them to the research community in an effort to find a cure for cystinosis. The CCIR is a central hub of information and is an essential tool for the global cystinosis research community. We are certain that the information provided by the patients will accelerate the research process and unite us in our quest to find a cure.

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



**CURE CYSTINOSIS**  
INTERNATIONAL REGISTRY

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



Thank you for your love, your generosity and for sustaining us.

## 2010 Spring Research Grants

Funded \$988,759

**Alan Davidson, PhD, Principal Investigator**  
Massachusetts General Hospital, Boston  
"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 and  
Ciaran Lee, Research Fellow**  
University College Cork, Ireland  
"Cystinosis Gene Repair"  
\$146,258 – 2-year grant

**Elena Levchenko, MD, PhD, Mentor and  
Joost Schoeber, PhD, Research Fellow**  
University Hospital Leuven, Belgium  
"Studying Podocyte Function in Nephropathic  
Cystinosis"  
\$150,000 – 2-year grant

**Miriam Britt Sach, MD, PhD,  
Principal Investigator**  
University of California, San Diego  
"In Vivo ATP Metabolism in Cystinosis Patients  
as Assessed by MR Spectroscopy"  
\$109,493 – 1-year grant

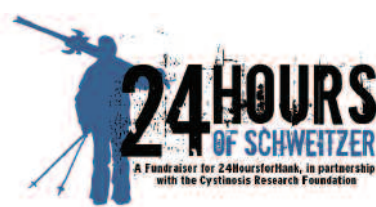
**Jennifer Simpson, MD, Principal Investigator  
James Jester, PhD, Co-Investigator**  
University of California, Irvine  
"Novel Treatment Modalities for Corneal Cystinosis"  
\$180,000 – 2-year grant

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



## Families United in the Quest for a Cure

- **Cycling for Cystinosis, Sandpoint, ID** – hosted and organized by Tricia and Brian Sturgis and the 24 Hours for Hank Foundation. This year 92 bike riders and 2 runners traveled 9,742 miles in honor of Hank Sturgis to raise more than \$33,000 for cystinosis research. Seven riders – including Greg Helbring, who lost five cousins in the early 1970's to cystinosis – rode over 300 miles. The next event, 24 Hours of Schweitzer will be held in April 2011. In just two years, the 24 Hours for Hank Foundation has raised more than \$200,000 to help find the cure! Sign up for the 24 Hour Ski event on the website [www.24hoursforhank.org](http://www.24hoursforhank.org)
- September 11–12, 2010 – **North Face 50 Mile Trail Run, Madison, WI** – Ruth Ann Ahnen completed the grueling race in honor of her daughter Katie who was diagnosed with cystinosis as a child. The day began at 5:00 am under cloudy skies with a 50 percent chance of thunderstorms. *As the race gets underway, thunder is heard, and the torrential downpour begins ...* despite the miserable weather Ruth Ann ran for more than 12 hours, raising more than \$3,800 for cystinosis research.
- **Denice and Mark Flerchinger** of Clarkston, WA, along with their family hosted the 2nd Annual Wine, Stein & Dine event, May 21, 2010 in honor of their daughter Tina, who has cystinosis. The event was a tremendous success and was highlighted by the attendance of the Helbling family of Moscow, ID who lost five of their ten children to cystinosis. The event raised \$44,000 for cystinosis research! [www.tinashopeforacure.org](http://www.tinashopeforacure.org)
- **Chrissy and Jason Grier** of Charlotte, NC, and their Hope for Holt Foundation have raised more than \$73,000 this year in honor of their son, Holt. The community support from their family and friends at the 2nd Annual Hearts for Holt fundraiser, Andrew Blair's Bistro dinner events, and the Kelly-McArdle Construction Golf Tournament has been amazing. [www.hopeforholt.org](http://www.hopeforholt.org)
- **Trevor and Jody Strauss** formed the Cystinosis Awareness & Research Effort (CARE), the first cystinosis organization in Canada, in honor of their daughter Gabbie. They have partnered with CRF to fund cystinosis research through multiple community fundraising events. CARE has raised more than \$70,000 this year. Learn more about them at [www.gabbieswish.com](http://www.gabbieswish.com) and [www.cystinosis.ca](http://www.cystinosis.ca)



## Welcome New Families

**Nicole and Brandon Cox** of Wake Forest, NC, recently established a foundation in honor of their daughter, Addison Rose, who was diagnosed with cystinosis when she was 17 months old. We are honored to have the Cox family as a CRF partner in our mutual quest to find a cure for cystinosis. After learning about Addison's story, Lenovo, Brandon's employer, selected CRF as the charity for their annual October Lenovo Cares Campaign. The Lenovo Cares Campaign allows employees to donate to CRF during the month of October. Thank you to the Cox Family and Lenovo for joining our quest for a cure. Visit their new website Addison's Angels, [www.addisonsangels.org](http://www.addisonsangels.org)

## Upcoming CRF Events – Save the Date

- **Saturday, February 12, 2011 ☆ Charlotte, NC**  
**Fourth Annual Hearts for Holt Event**, in honor of Holt Grier. A lively fun-filled evening including dinner, entertainment, and live and silent auction, is planned at the beautiful Charlotte Country Club benefiting the Hope for Holt Foundation. [www.hopeforholt.org](http://www.hopeforholt.org)
- **Thursday, March 24, 2011 ☆ Sacramento, CA**  
**The Association of Commercial Real Estate (ACRE) hosts a Charity Benefit/Broker of the Year Awards dinner and fundraiser.** Because of the dedicated efforts of Teresa and Kevin Partington, ACRE has chosen the *Jenna and Patrick Foundation of Hope* as its charity recipient for the 2011 and 2012 Broker of the Year events. [www.jennaandpatrick.org](http://www.jennaandpatrick.org)
- **Friday, April 1 – Saturday 2, 2011 ☆ Sandpoint, ID**  
**Third Annual 24 Hours of Schweitzer Ski Event** in honor of Henry Sturgis. This marathon event is for all level of skiers, snowboarders and those who love a challenge. [www.24hoursforhank.org/events.html](http://www.24hoursforhank.org/events.html)
- **Friday, May 20 – Saturday 21, 2011 ☆ Newport Beach, CA**  
**CRF Day of Hope** family conference will bring together families and leading cystinosis researchers and scientists. The researchers will present their research updates and answer questions about cystinosis. This is a unique opportunity for families and researchers to share information and focus on their mutual mission – to cure cystinosis.
- **Saturday, May 21, 2011 ☆ Newport Beach, CA**  
**The 10th Annual Natalie's Wish Event** promises to be an evening of inspirational stories and a celebration of the extraordinary progress CRF-funded scientists and researchers have accomplished since 2003. [www.cystinosisresearch.org](http://www.cystinosisresearch.org)



Mark Your Calendars –

**Fourth Annual Natalie's Wish Four A Cure** Golf Tournament to be held at the beautiful Santa Ana Country Club, on **Monday, November 14, 2011.**

100 percent of all donations go directly to cystinosis research. Your gift is tax deductible. The Cystinosis Research Foundation is a non-profit, tax-exempt entity pursuant to Section 501(c)3. Federal Tax ID # 32-0067668



## CYSTINOSIS RESEARCH FOUNDATION

18802 Bardeen Avenue ■ Irvine, CA 92612-1521

(949) 223-7610 ■ [www.cystinosisresearch.org](http://www.cystinosisresearch.org)

