

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

# CYSTINOSIS RESEARCH FOUNDATION

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

Dear Friends and Family,

*As the holidays draw close, our family has so much to be thankful for. This year has been one of hope and scientific progress. And now, more than ever, we are amazed and eternally grateful for the generosity you have all displayed.*

*We have come so far in the last few years and we have much to be thankful for...*

- Our annual *Natalie's Wish* event raised \$1.5 million for Cystinosis research thanks to the generosity of our family and friends. In just four years, we have funded 17 research studies totaling over \$2.7 million.
- In October we announced a \$1.2 million global call for Cystinosis research proposals. After review by our Scientific Research Board, we will announce approved studies by the end of this year.
- The Cystinosis Research Foundation launched the first post-doctoral Cystinosis Research Fellowship Program to encourage young investigators to establish careers in Cystinosis research. Fellowship awards will be announced by the end of this year.
- The Cystinosis Research Foundation was honored to be chosen by Traditional Jewelers of Newport Beach and *Riviera Magazine* as the recipient charity for the **Diamond and Jewelry Holiday Extravaganza** held on **Thursday, November 9, 2006**. A portion of the evenings proceeds were donated to CRF.
- The CRF will hold its inaugural **Golf Tournament for Cystinosis** at the Coto de Caza Golf Club on **December 3, 2007**. The event will be chaired by Renee Carter. Sponsors and volunteers are needed and welcome!
- The second phase of the clinical, **slow-release medication study is underway** at the UCSD Medical Center. To date, four children have been studied. Although it is early, the initial data looks promising.

In October, Natalie and three other brave children with Cystinosis volunteered for the second phase of the research study to find a slow-release form of the current and only medication available to treat Cystinosis. In February, 2006, the CRF issued a grant to Dr. Dohil entitled, "A Study to Evaluate Enteric-Coated Cysteamine Therapy in Patients with Cystinosis." If successful, the study will dramatically change the life of children with Cystinosis. Currently, medication must be taken every 6 hours, every day just to stay alive. **There are no days off for children and young adults with Cystinosis. They have never slept through the night because of the grueling medication schedule.**

The study required children to stay at the UCSD Medical Center for one week. During that time the children were subjected to many tests including blood chemistry tests, white cell cystine and cysteamine level tests and urinalysis. The children took doses of the current medicine and then of the new coated medicine. After taking the medications, blood was drawn and analyzed. On certain days, it was drawn every 15 minutes. A line was inserted to draw blood which helped ease the pain, anxiety and needle sticks. Throughout the week, more than 120 blood samples were taken!

It was a long and stressful week for the children and their parents. Hospital visits for children with Cystinosis are not uncommon, however, being part of a research study seeking better treatments reminds us of how sick our children are and how desperately we want to save their lives.

The children were sent home with a 30-day supply of the coated 12-hour medicine. Each week for four weeks after the study, blood was drawn to measure the effects of the coated medicine. **For Natalie, it was the first time in 15 years that she had slept through the night, and the first time she did not have to bring her medicine to school.** It has been a bittersweet month because we are back to the 6-hour schedule until the study is complete. **It has however, been a remarkable experience for Natalie to know what a full night's sleep is like. She has tasted what we hope the future will be like for her and the other children with Cystinosis. She has lived a touch of heaven for the past 30 days. We are grateful we had the opportunity to participate in the study.**

Natalie begged me to ask Dr. Dohil if she could stay on the 12-hour schedule and the coated medicine. She knew the answer would be "no" and although she is disappointed, she understands that we must wait for completion of the trial and analysis of the data from all of the children volunteers before we know if the coated medicine is effective.

Although it is hard to go back to waking Natalie up twice a night and to taking her medicine at school, we have been very fortunate to know what her life and the lives of the other children with Cystinosis might be like in the near future.

We have the gift of hope and that will sustain us. We pray that the results will be positive and that it won't be long before the coated medicine will be a reality.

*We are grateful to each and every one of you for making this clinical trial a reality. You have given the entire Cystinosis community the greatest gift of all – hope.*

*Jeff, Natalie, Nancy and Alexandra Stack*



www.natalieswish.org

**SAVE THE DATE!**

**Thursday, May 31, 2007**

**Sixth Annual Natalie's Wish Event**

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## 2006 Research Studies

- **Ranjan Dohil, MD;** University of California, San Diego; whose cysteamine absorption discoveries were derived from previous CRF-funded research and published in the June 2006 issue of "The Journal of Pediatrics," received \$253,685 for a one-year Phase II research study to evaluate enteric-coated cysteamine in patients with Cystinosis. The purpose is to determine if enteric-coated cysteamine will last longer in the body than the current form. If so, it might be possible to take the medication every 12 hours instead of every 6 hours.
- **Corinne Antignac, MD, PhD;** Necker Hôpital, Paris, France; received \$85,000 for a one-year study on the characterization of Cystinosis intracellular trafficking. The project, a continuation of the project funded in 2005, is aimed at understanding the exact role of cystinosin (the Cystinosis gene product discovered in 1998) in the cells and why its alteration leads to development of Cystinosis.
- **Stephanie Cherqui, MD and Daniel Salomon, MD;** Scripps Research Institute, La Jolla; received \$709,170 for a three-year study to research the treatment of Cystinosis nephropathy using genetically modified adult stem cells. The objective is to test the hypothesis that transplantation of autologous adult stem cells expressing a functional CTNS gene (the defective gene) can be used to treat and prevent renal defects of Cystinosis.
- **Angela Ballantyne, PhD, and Amy Spilkin, PhD;** University of California, San Diego; received \$213,527 for a two-year study to examine the academic functioning in children with Cystinosis using a comprehensive achievement battery. The study will serve as the basis for future application of intervention strategies that may lead to greater academic success for children with Cystinosis.
- **E.N. Levchenko, MD, PhD;** University Medical Centre St. Radboud Nijmegen in The Netherlands received \$72,423 for a one-year study on the pathogenesis of interstitial renal damage leading to renal failure in Cystinosis. End-stage renal disease is the leading cause of morbidity in Cystinosis patients.
- **Jerry Schneider, MD and Bruce Barshop, MD, PhD;** University of California, San Diego; received \$118,400 for a two-year lease and maintenance of a tandem mass spectrometer needed for Cystinosis research.

## Scientific Review Board

*We are pleased to announce that Julie Ingelfinger, MD has agreed to join our Scientific Review Board. Dr. Ingelfinger is Deputy Editor of the New England Journal of Medicine, Professor of Pediatrics for Harvard Medical School, and Senior Consultant for the Pediatric Nephrology Division at Mass General Hospital.*

*We welcome Dr. Ingelfinger and want to thank all of the SRB members for their support and commitment to improving the lives of our children.*

### Chairperson

**Jerry A. Schneider, MD**  
Research Professor of Pediatrics  
Dean for Academic Affairs Emeritus (RTAD)  
University of California, San Diego

### Board Members

**Corinne Antignac, MD, PhD**  
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Director of Inserm Research Unit U574  
Hôpital Necker-Enfants Maades  
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**William Rizzo, MD**  
Department of Pediatrics  
Chief, Inherited Metabolic Diseases  
University of Nebraska Medical Center

**William van't Hoff, BSc (Hons), MD, FRCP, FRCPC**  
Consultant Pediatric Nephrologist  
Great Ormond Street Hospital  
London, England

## 2006 Autumn Call for Funding Proposals

**Research Proposals:** The Cystinosis Research Foundation announced its second 2006 call for research proposals in early October. The CRF is prepared to fund proposals to improve the immediate care of children and young adults with Cystinosis and to develop new understandings and treatment of Cystinosis to help these children in the future. The Foundation has more than \$1.2 million in research funds available. The number of awards and their value will depend on the number of outstanding proposals received in November and the funds available at the time.

**Post-Doctoral Fellowships:** The CRF is establishing the first post-doctoral research fellowship program in the United States to encourage young investigators to establish careers in Cystinosis research. Fellows will be funded for 2-3 years to a maximum of \$75,000 per year.

Proposals are reviewed by our Scientific Review Board who advise the CRF on the scientific merit of each proposal. The CRF balances the eventual funding to support clinical and bench research and fellowships. Research grants will be announced at the end of 2006. Check [www.natalieswish.org](http://www.natalieswish.org) for updates.

# CYSTINOSIS RESEARCH FOUNDATION

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100 percent of all donations go directly to Cystinosis research. Your gift is tax deductible.

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