

Natalie's Wish

*Star light, star bright,
First star I see tonight
I wish I may, I wish I might
Have the wish, I wish tonight.*

FOR FRIENDS OF NATALIE STACK AND SUPPORTERS OF THE CYSTINOSIS RESEARCH FOUNDATION ★ FEBRUARY 2007

A Note from Natalie

I started my sophomore year at Sage Hill this past fall. It has been an exciting and successful year so far. I am on the JV basketball team again. This season is much better than last season because we have won a few games and there are more players on the team. I have also joined a school club that helps poor families around the world.



I was part of Dr. Dohil's slow-release study. I was in the hospital for a week taking the old medication and then trying the new, slow-release medication. The nurses measured my blood levels everyday sometimes every fifteen minutes! When I first took the slow-release medicine the double dose was very strong and too much for me to handle. I smelled like the medicine. My mom told the doctor she could smell it from outside the room. My stomach was also queasy and I was very sleepy. One day when I left for a break, I fainted in the elevator. After that, Dr. Dohil gave me half of the dose.

With Dr. Dohil's help I was able to remain on the slow-release medicine for three additional months and it has changed my life. Taking medicine twice a day is so much better than taking it every six hours. I do not have to bring medicine to school and I have been sleeping more soundly because I do not have to wake up at midnight and five in the morning to take my medicine. With the old medicine, my stomach hurt all day long but it now feels more normal than ever. With the old medicine, my stomach was bloated at least once or twice a day and if it wasn't bloated, it hurt.

Soon I will have to go back to the old way of taking the medicine. I am just glad that I was able to try this medicine to know what it feels like to have a more normal life and to be able to sleep through the night. I hope that the rest of the study goes well.

Thank you to my friends, family and doctors for all your support. The money raised in only a few years is beyond belief! You have made a difference in my life and in the lives of the other children with Cystinosis.

Thank You! ♥ Natalie Stack



Natalie with Kevin Sharp and her sister Alex Stack at last year's Natalie's Wish event.

Dear Friends and Family

In 2003, when we established the Cystinosis Research Foundation (CRF) our goal was to aggressively pursue new and better treatments for Cystinosis and ultimately to find a cure for the disease. With your help we are moving rapidly in that direction.

Only four short years ago Cystinosis research was minimally and sporadically funded. Clinical research and multi-year research studies were rare, and it was difficult to attract new researchers. Today, because of your commitment, CRF is the largest non-profit funder of Cystinosis research in the world.

There is hope for those who suffer from Cystinosis because of the ongoing research you have funded.

In 2006 alone, CRF raised more than \$1.6 million. Overall, the Foundation has raised more than \$4.0 million and has committed and funded more than \$3.5 million for Cystinosis Research. CRF is now funding 11 research studies and three research fellows. Our researchers are working in four countries around the world and are collaborating to ensure the sharing of ideas and energy.

This mailing includes our **Cystinosis Research Foundation Science Report**. In the report, you can learn about recently funded research, our research fellows and review progress reports on previously funded research.

continued on page two

CYSTINOSIS RESEARCH FOUNDATION LAUNCHES RESEARCH FELLOWSHIP PROGRAM

The Cystinosis Research Foundation has established the first Research Fellowship Program in the United States to encourage young investigators to establish careers in Cystinosis research.

Our vision is to promote and foster innovation, collaboration and forward-thinking research among qualified young scientists at leading institutions and research centers around the world.

Our goal is to serve the Cystinosis community as the foremost resource to fund life-changing new treatments and support clinical programs. We are proud to announce we have awarded three grants in 2006:

Eric Moses, PhD, Mentor

Katy Freed, PhD, Research Fellow

Southwest Foundation for Biomedical Research, San Antonio, Texas

“Complex Genetic Approaches to Monogenic Disease: Genomic and Transcriptomic Dissection of Normal Expression of CTNS, the Gene Involved in Nephropathic Cystinosis”

\$148,535 – 2-year fellowship

Vasiliki Kalatzis, PhD, Mentor

Claire Hippert, Pre-doctoral Research Fellow

Institut Génétique Moléculaire Montpellier, Montpellier, France

“Gene Transfer Studies for Cystinosis”

\$168,290 – 2-year fellowship

Holger Willenbring, MD, Mentor

Kentaro Okamura, PhD, Research Fellow

University of California, San Francisco

“Targeted Cell Fusion for the Correction of Tubulopathy Due to Cystinosis”

\$84,530 – 2-year fellowship



Dear Friends and Family *continued from front page*

One of the most exciting accomplishments of last year was the creation of the Cystinosis Research Foundation Research Fellowship Program – the first such program ever established. The purpose is to encourage

bright, young scientists to enter the field of Cystinosis research. We are pleased to announce that we have awarded two-year grants to three new scientists who have demonstrated a commitment to Cystinosis research.

We are also excited to report on the second phase of Dr. Ranjan Dohil's clinical trial. To date, five children with Cystinosis have volunteered for the demanding one-month study that involved testing an enteric-coated form of the current medication. The enteric-coated medication is administered every twelve hours, versus the current, every-six hour protocol. The aim of the study is to test the enteric-coated Cystagon™ to determine whether it is absorbed better and released more slowly thus reducing the drug's constant and painful side effects.

The study has produced tangible, better-than-expected preliminary results, and Dr. Dohil and his colleagues are extremely encouraged. In fact, because of the excellent, early results, two children, Natalie being one of them, have continued on the therapy for three additional months.

For 15 years Natalie has taken her medication every six hours of every day including twice each night. She has never had a full night's sleep until she was part of the enteric-coated study. We are grateful to Dr. Dohil and Dr. Schneider for their dedication to Cystinosis research. And we are proud of the five courageous children who volunteered to be part of this difficult study.

Dr. Dohil will continue the study with additional volunteers. He will work

to establish the correct formulation, while minimizing gastrointestinal symptoms. We anticipate the new data will continue to be positive as we pursue a drug company's support to move this formulation through the FDA approval process.

Every week I hear from families whose children are suffering. They are thankful for the work CRF is doing, and they hope that soon their children will be on the slow-release medication, which will significantly improve their children's lives.

In 2003, when CRF was established, one of our goals was to improve the quality of life for children with Cystinosis. Finding more effective treatments was the key to that goal. It was impossible then to imagine daily life without the onerous six-hour medicine schedule. Today, we are on the brink of a better treatment. Your prayers, financial support and commitment have brought us closer than ever to extraordinary changes in the daily lives of children with Cystinosis.

We thank you for your commitment. We are grateful to those who are dedicated to making our upcoming Natalie's Wish event a success. We are especially thankful for the tireless efforts of Zoe Solsby and Marylyn Milburn whose daily commitment and energy make the CRF so successful.

We hope you will join us for this year's event on Friday, June 1 at the Balboa Bay Club. The evening promises to be emotional and inspirational with NFL football legend Jim Kelly sharing the story of his son's courageous battle with a rare disease. It is a story of hope and heartbreak you will not want to miss.

Nancy and Jeff Stack



NFL legend Jim Kelly

SIXTH ANNUAL NATALIE'S WISH

 FEATURING NFL LEGEND JIM KELLY

FOURTH AND GOAL

The courage to try. The will to win.

FRIDAY, JUNE 1 BALBOA BAY CLUB

The lives of children and young adults with Cystinosis continues to be a daily struggle. Because of your help over the last five years, there is hope and progress. The ultimate victory – a cure for Cystinosis – remains our steadfast goal. As we plan this year's event, we are on the verge of an extraordinary breakthrough in treatment that brings us closer to our goal of improving the quality of life for those with Cystinosis.

We invite you to join us in supporting this year's Natalie's Wish event featuring NFL legend Jim Kelly who will share his professional story of triumph and his family's story of courage, hope and heartbreak.

This year's event will also feature the always inspiring Cystinosis video, Orange County's most sensational auction and the highly anticipated *Fund-A-Cure*. And there will be a surprise or two to make this year's event our best ever.

For information call Zoe Solsby at 949.223.7610 or visit www.natalieswish.org.



Diamond Extravaganza Yields \$35,000 for CRF

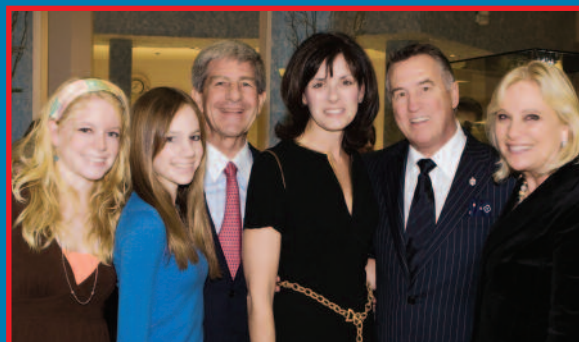
Natalie's Wish and the Cystinosis Research Foundation was the beneficiary of a \$35,000 donation from the Traditional Jewelers Diamond and Jewelry Extravaganza in Newport Beach in late November 2006. More than 300 people attended the event that included exceptional jewelry designs and the display of a rare, pink 10-carat diamond.

"We are so very grateful for this major donation and to have been selected as the charity of choice by Traditional Jewelers for this annual event. Lula and Marion Halfacre have embraced the foundation and are major contributors to our research efforts" said Nancy Stack.

Traditional Jewelers, one of Southern California's leading sellers of fine jewelry, committed a percentage of the evening's sales to benefit Natalie's Wish. Lula and Marion Halfacre, owners of Traditional Jewelers, have a long history of supporting local causes. Each year they select one charity as the beneficiary of the annual Diamond and Jewelry Extravaganza. Dozens of jewelry artisans from around the world displayed their creations – some of the most exquisite jewelry available anywhere.

Just in time for the holidays, pre-holiday shoppers were enthusiastic and supportive of the event, especially Lynette and Michael K. Hayde, chief executive officer of Western National Group. By virtue of their major jewelry purchases, they won a five-day trip to Pueblo Bonito Sunset Beach Resort & Spa in Cabo San Lucas. The Haydes graciously donated the trip to the live auction for the upcoming Sixth Annual Natalie's Wish Event, Friday, June 1, 2007 at the Balboa Bay Club.

We are thankful to everyone who was able to attend and wish to express our gratitude to Traditional Jewelers for helping Natalie's wish come true.



Natalie, Alex, Jeff and Nancy Stack with Lula and Marion Halfacre

Tiki Boat FUNDRAISER

The 2nd Annual Harbor Day School "Class of 2005" Reunion raised more than \$4,000 for Natalie's Wish and the Cystinosis Research Foundation. The "Tiki" harbor cruise was held on May 26, 2006 and was hosted by Natalie's friends Kelsey Valley, Shelby Searles, Caroline Kelter and Jenny Madden. The boat toured Newport harbor for several hours while guests dined and danced to the sounds of a DJ.

The event, which was originally a birthday party cruise, has raised a total of \$10,614. The girls are honored to help bring Natalie's wish, "to have my disease go away forever" closer to reality.

We are grateful to Natalie's friends for their efforts to help Natalie and others with Cystinosis.



Shelby Searles, Kelsey Valley, Natalie Stack, Jenny Madden, Caroline Kelter

You Can Help FUND A CURE

There are many ways you can help children with Cystinosis. One of the best ways is to hold a fundraiser for CRF – the possibilities are endless!

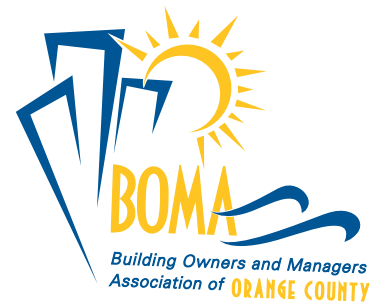
Here are just a few suggestions:

- Instead of gifts at your birthday or anniversary party, ask guests to bring donations
- Run a bake sale at your school or church
- Hold a car wash or dog wash
- Host a garage sale
- Gather pledges for a run, walk or bike to benefit Natalie's Wish

We'll provide all the support you need – a supply of newsletters and brochures, suggestions for fundraising and lots of moral support! Of course you are free to develop one of your own creative ideas into a fundraiser. Please contact the CRF office for assistance, 949.223.7610

www.natalieswish.org

BOMA OC SCORES BIG FOR NATALIE'S WISH



The Cystinosis Research Foundation has received a donation of \$18,515 from the Orange County Chapter of the Building Owners & Managers Association.

The money was raised at the auction following the sold-out golf tournament and dinner August 2 at the Talega Golf Club in San Clemente.

During the past three years BOMA Orange County has given \$40,119 to Natalie's Wish. "BOMA has contributed more to CRF than any other industry group. Their generous contribution has helped us fund important scientific and medical studies – studies that are producing very promising results in treating this deadly and rare disease, which mostly affects children," said Nancy Stack.

"Many other charities we looked at receive public and private financing. We were intrigued with CRF because of its local ties and it is completely privately funded," said Mike Raring, co-chairman of the BOMA golf tournament.

"We are gratified to play a part in advancing scientific research that is producing results with such promise," Raring said. "The Stacks are such great, everyday people. And they've already made tremendous progress. You never know what you're going to unlock when you find the key to that door. Hopefully we can make Natalie's wish come true."

Thanks to BOMA's Golf Committee:

Mike Raring, *tournament co-chairman*
AAA Electrical & Communications Inc.
Jon Schneider, *tournament co-chairman*
Specialty Uniform Services

Kim Bangs
AAA Electrical & Communications Inc.
Dorothy Bisaha, *SERVPRO of Tustin*
Judy Bowers, *Cox Business Services*
Bob Brans, *Structural Materials Co.*
Dana Casassa
EMCOR/Mesa Energy Systems Inc.
Dale Fedele, *Ampower*
Seth Foster, *HSG Inc./Bird Busters South*
Kathleen Gillett
The Irvine Company Office Properties
Lysa Horn, *AlliedBarton Security Services*
Terri Houllis
Day-Lite Maintenance Company Inc.
Robin Jochims, *Executive Director*
of BOMA's Orange County Chapter
Jeff Koscher
Advanced Restoration Specialists Inc.
Sepp Maier, *The Lighting Company*
Lisa Miller
CarrAmerica Realty Corporation
Monique Mora
Statewide Acquisitions Corp.
Betty Pickett, *Universal Protection Service*
Eric Sorensen, *Able Engineering Services*
Brett Wells
Frazee Paint & Wallcovering

SEARCHING FOR A CURE

GoodSearch

The Cystinosis Research Foundation is now listed on GoodSearch.com, a search engine that donates about a penny per search, to the charities its users designate. Every time you use GoodSearch.com (instead of your current search engine, such as Google) money goes to CRF! The site is powered by Yahoo, so you'll get the same quality search results that you're used to.

To find out more visit: www.goodsearch.com

Be sure to type in Cystinosis Research Foundation in the "my charity" box. It takes less than 20 seconds to set up! And remember to tell your friends. If 1,000 people search twice a day for a year, CRF could earn \$7,300!



An Unusual Christmas Gift

We will never forget December 18, 2003.

We wanted one Christmas gift.

It was all we asked for.

Finally with Christmas a week away, we got the news of what was making our 2 1/2-year-old daughter Alexis gravely ill.

Cystinosis?

Our first thought was, "What was it?"

We were told that it is a rare, genetic, metabolic disease that causes kidney failure, blindness, muscle-wasting, bone disease and many other complications.

We were scared— afraid of what it all meant. Was it the needle-in-the-haystack diagnosis we were desperate to find?

There were so many emotions.

Born June 16, 2001 Alexis entered the world healthy, or so we thought.

By 12 months of age she had stopped eating, stopped growing, drank more than twelve 8 oz cups of water, and urinated as many times as she drank, vomited and gagged all day and night. We knew she was dying but the doctor's insinuated we were spoiling her.

By two years old she began intervention therapies in our home and began seeing three specialists. We switched pediatricians and began seeing a nephrologist who finally diagnosed Alexis. By this time we had a son, Evan who was two months old, born October 22, 2003. The doctor decided to test him too. At three months, we learned he too had Cystinosis. Dealing with one child with a chronic illness was devastating but two, almost impossible. And so began the 1 1/2-year struggle to stabilize the kids, ensuring that all six of their combined medicines were given around the clock.

Today we look back on the last three years and are grateful for many things that Cystinosis has brought into our lives. Alexis is 5 1/2 years old and in kindergarten. She loves school, coloring and her friends. She oozes empathy when others are sick or hurt. Her biggest struggles are fatigue and understanding why she has to take so many medicines. She hates being pulled twice in her nearly three-hour-day from school to take one of the eight medicines and she prays that it will all go away someday. She knows she has little control in her life with Cystinosis, so she does everything she can to get it back outside of it.

Evan is three and because of early diagnosis and treatment he is almost as tall as Alexis and his kidneys are healthy. He currently takes four medications around the clock but doesn't complain. He is a typical boy, full of life and energy. He was born with a smile on his face. He struggles with a dominant sister and sitting still. He loves to swim, dance and play guitar. He has taught Alexis how to swallow pills, overcome fear and love life despite feeling ill.

As their parents, at a minimum we only hope they will know what a full night sleep is like. Sleep deprivation has taken its toll on our family. We have a deep desire for balance in our lives. Our commitments are minimal. We still struggle with living with Cystinosis. We will always fear if we will out live our children or what kind of quality of life they will have as they grow up. We have met many extraordinary families, doctors and friends through this journey. Cystinosis has made us more selfless and turned our focus outward meeting the needs of others.

So it is this Christmas, three years later that we hope to be a blessing to those who have blessed us.

In Him,

Brett, Brittney, Alexis and Evan Le Beau

Dealing with one child with a chronic illness was devastating but two, almost impossible. And so began the struggle to stabilize the kids, ensuring that all six of their combined medicines were given around the clock.





Friends and Families

Sophia Towery's Story

By Debra Towery, Sophia's mother

My daughter Sophia Antonia Towery is a first-grade seven-year-old. We live in Sumter, South Carolina. I learned Sophia had Cystinosis in October 2000 when she was 16 months old. She also was diagnosed with Fanconi Syndrome and rickets. The pediatric nephrologists at The Medical University of South Carolina had dealt with Cystinosis patients before and knew the diagnosis after one look at Sophia. They said her blonde hair and blue-eyed fair complexion were a give away.

Sophia began the vigorous regimen of medication, but she was unable to take the volumes of medication by mouth without vomiting. She had a gastronomy tube placed in her stomach to provide the medication and nutrition she needed. Sophia finally learned to swallow pills and no longer needs the "g-tube."

Sophia currently takes 33–35 pills a day and eye drops 8–10 times a day for photosensitivity.



Sophia was also on growth hormone for four years which involved an injection every night into her stomach. She is currently off of this medication because the insurance (again) says it is not a medical necessity.

Sophia also has problems with her heart, scoliosis and leg pain from being "bowed" by rickets. In October, she underwent surgery on her left leg. She was homebound for three months with pins and an external fixator in her leg, and a metal plate and screws in her ankle.

On January 11, the doctors thought that Sophia's leg had healed and they took the pins out and told her not to put any weight on the leg for one month. No sooner did we have her home when she screamed in excruciating pain. We returned to the doctor and discovered her femur bone was broken! Sophia went back into the hospital where doctors made new incisions, placed all six pins and the external fixator back in her leg. She cannot attend school for another two to three months and is in a wheelchair. She is most upset about not being able to swim, and she is anxious to walk again and see her friends at school.

Sophia is a strong-spirited child who deals with adversity on a daily basis. She brings a smile to the face of everyone she meets. Cystinosis has brought many hardships for Sophia and our family, but we have learned to enjoy everyday we are given.

Alex Weaver

By Angie Weaver, Alex's mother



Alex was diagnosed with Cystinosis when he was 11 months old. Initially, he would not eat and got so sick he started to lose weight. One weekend, when he was especially sick, I took him to the doctor's office where they ran a few tests. We were immediately sent to the emergency room, and he was admitted to the hospital. We spent three nights and four days there. After being pounded with questions from various doctors, a pediatric nephrologist told us he thought he knew what was wrong.

Our simplistic lives changed at that moment. I remember it like it was yesterday. The doctor ordered more blood work, but he let us go home after warning us not to look up anything on the internet because he didn't want us to see anything that might scare us. That statement, in itself, terrified us. He told us that this disease would eventually shut down Alex's vital organs. Then, the denial started. I thought, there was no way Alex was going to have this disease. We were not going to watch our child die. We were fortunate that Alex was diagnosed so quickly but, on the other hand, the doctor was not very positive about his future. He advised us that Alex would probably need a feeding tube and would be in and out of the hospital. We did a lot of research and found several support groups, which enabled us to talk with other families who had children with this disease. We even spoke to Dr. William Gahl, one of the world's leading experts on Cystinosis. He explained that this illness wasn't a death sentence, and that Alex would be able to live a relatively normal life, but he would need to take many different medications forever or until a cure was discovered.

The doctor warned us not to look up anything on the internet because he didn't want us to see anything that might scare us. That statement, in itself, terrified us.

Alex is now four years old and doing excellent. He is in the 75th percentile for his height and weight. You would never know he has any medical issues. He currently takes six different medications. Even now, he sometimes chokes and spits the medicine up. I know I'm doing the right thing by making him take the medication, but it is emotionally challenging watching and listening to him cry. We have begun transitioning him to swallowing capsules.

It is amazing how much life can change in a matter of minutes. We have learned not to take things for granted. From the beginning, we have dealt with Alex's illness in a proactive way, and feel that has made an incredible difference for him and our family. We were fortunate that he did not have to get a feeding tube, and he has not been in the hospital since his diagnosis. We will continue to pray for a cure, and in the mean time, we will enjoy the blessings we have in our lives.

I cannot begin to express how thankful we are for the support groups that are available. You are the reason Alex is thriving today. You give us hope. Thank you!

MEET THE CRF SCIENTIFIC REVIEW BOARD

JERRY A. SCHNEIDER, MD

Dr. Schneider received his MD degree from Northwestern University Medical School. He did his pediatric internship and residency at the Johns Hopkins Hospital. From 1965 to 1967 he began to study Cystinosis as a Clinical Associate at the National Institutes of Health (NIH). This was followed by an additional two years at the NIH and one year at the Centre de Génétique Moléculaire in Gif-sur-Yvette, France studying biochemistry and genetics. He joined the Department of Pediatrics at the University of California, San Diego in 1969. He was named the Benard L. Maas Chair of Inherited Metabolic Diseases in 1986 and Dean for Academic Affairs in 1995.



Dr. Schneider spent two sabbatical years in London, England at the Imperial Cancer Research Fund Laboratories, the first as a Guggenheim Fellow and the second as a Fogarty Senior International Fellow. He has been an Established Investigator of the American Heart Society and is a member of numerous societies. In 1995 he was awarded the FDA Commissioner's Special Citation for his work in developing cysteamine as a treatment for Cystinosis. He has written over 150 publications, most dealing with Cystinosis. Dr. Schneider retired from his tenured position at the University of California, San Diego in 2004. The University recalled him as a Research Professor, allowing him to continue his Cystinosis-related studies.

CORINNE ANTIGNAC, MD, PHD

Corinne Antignac's laboratory research programs are mainly devoted to the identification of genes involved in hereditary renal diseases and to the characterisation of the proteins encoded by these genes. After her training as a Paediatric Nephrologist in Paris, France, Corinne Antignac obtained a PhD in Human Genetics at Paris 6 – Pierre et Marie Curie University and set up her research group at Inserm (French National Institute of Health and Medical Research) in Necker Hospital in 1990. Since then, she became Prof. in Human Genetics at Paris 5 – René Descartes University in September 2001 and is Director of the Inserm group U574 (4 permanent Inserm researchers, 5 post-doctoral associates, 5 graduate students and 7 research technicians) since January 2003. She is member of several national and international societies (Société de Néphrologie Pédiatrique, Société de Néphrologie, European Society for Paediatric Nephrology, International Society of Nephrology, American Society of Nephrology) and numerous national scientific boards.



Corinne Antignac's main results concern the identification, by positional cloning, of four genes, NPHP1, NPHP4, CTNS and NPHS2 underlying hereditary kidney disorders, nephronophthisis, Cystinosis and steroid-resistant nephrotic syndrome. In all cases, the genes appeared to encode new proteins of unknown functions, which have been (or still are) characterised in the laboratory. In particular, C. Antignac's group has shown that cystinosis, the CTNS gene product, is a lysosomal membrane protein and a cystine – proton symporter. Animal models, mostly by inactivation of the homologous gene in mouse, have been successfully generated, allowing better analysis of the disease phenotype and the future use of these models for testing emerging therapies. The group has also been involved in identifying genes by candidate gene approaches and in phenotype/genotype correlation in other hereditary renal disorders such as Bartter and Alport syndromes, with a particular emphasis on Alport syndrome and diffuse oesophageal leiomyomatosis. Altogether, these works lead to the publication of more than 120 original articles.

The Scientific Review Board is composed of leading Cystinosis scientists and experts from around the world who have dedicated numerous volunteer hours to the Cystinosis Research Foundation. The members are actively involved in the grant review process, evaluating and analyzing all research proposals submitted to the Cystinosis Research Foundation. The Board advises the Foundation on the scientific merit of each proposal.

We are indebted to our board members for their breadth of knowledge, their guidance and commitment to helping our children.

Scientific Review Board

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MEET THE CRF SCIENTIFIC REVIEW BOARD

MICHEL BAUM, MD



Dr. Michel Baum received his BS and MD degrees from the University of California at Los Angeles. He did his internship and residency at Yale University School

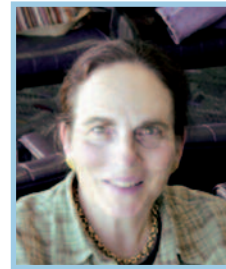
of Medicine. His nephrology fellowship was performed at the University of California at San Francisco, where he was a research fellow at the Cardiovascular Research Institute.

Since completion of his fellowship he has been at The University of Texas Southwestern Medical Center and Children's Medical Center of Dallas. He is currently Professor of Pediatrics and Internal Medicine and Director of Pediatric Nephrology. He also holds the Sara M. and Charles E. Seay Chair in Pediatric Research. He is a member of a number of national societies including the Association of American Physicians, the American Society for Clinical Investigation, the Society for Pediatric Research, the American Pediatric Society and the American Society of Nephrology and Pediatric Nephrology. He has served as a member of the General Medicine B Study Section for four years and on numerous national committees. He is Co-Editor of *Pediatric Nephrology*.

Dr. Baum's research primarily examines the maturational changes that occur in renal tubular transport during development. He has described many changes in transporters that explain the physiologic difference between the neonatal kidney and the adult kidney. His other research interests include the pathophysiology and pathogenesis of hereditary defects in tubular transport such as X-linked hypophosphatemia and Cystinosis. He has also conducted many studies examining how hormones regulate tubular transport.

Dr. Baum's research focuses on patients with metabolic bone disease including X-linked hypophosphatemia. His research is funded by the National Institutes of Health. He has over 100 publications.

JULIE R. INGELFINGER, MD



Dr. Ingelfinger is Senior Consultant in Pediatric Nephrology, MassGeneral Hospital for Children of Massachusetts General Hospital and Professor of Pediatrics, Harvard Medical School. She has also been a member of numerous journal editorial boards. Dr. Ingelfinger received her undergraduate degree from Harvard University [Radcliffe College] and her MD from Albert Einstein College of Medicine.

A pediatric nephrologist, Dr. Ingelfinger has focused clinical care and research as well as basic laboratory studies in the areas of pediatric hypertension and nephrology. She has published over 230 papers and review articles and has authored or edited nine books. Dr. Ingelfinger is a member of the American Pediatric Society, the Society of Pediatric Research, The American Society of Nephrology, The American Society of Pediatric Nephrology and other national and international societies, and she has served as president of The American Society of Pediatric Nephrology.

She has received numerous grants, awards and prizes. Dr. Ingelfinger is involved in developing reviews and editing original articles at *The New England Journal of Medicine*, focusing on the Medical Progress series and on original articles in her areas of expertise, pediatrics, pediatric nephrology and basic science. She has lectured nationally and internationally on nephrology, hypertension and issues that involve the caring for children.

WILLIAM RIZZO, MD



Dr. William Rizzo is Professor of Pediatrics at the University of Nebraska Medical Center in Omaha and Director of the Hobart Wiltse Center for the Study of Inherited Metabolic Disorders. He obtained a BA from Northwestern University in 1972 and a MD from the University of Illinois in 1977. He did an internship and residency in Pediatrics at Johns Hopkins University and subsequently completed fellowship training in Medical Genetics at the National Institutes of Health (NIH) in 1982. During this time, he took care of many of the Cystinosis patients followed at the NIH.

Dr. Rizzo joined the Medical College of Virginia in 1983 as Assistant Professor and was promoted to Professor in 1993. While there, he developed a new dietary approach for treatment of adrenoleukodystrophy, which later became known as Lorenzo's Oil after the popularized film by that same name. In 1988, Dr. Rizzo discovered the enzyme defect that causes Sjögren-Larsson syndrome, a genetic disease that affects the skin and brain of children. In 1995, he spent a sabbatical year studying molecular biology at the NIH and cloned the gene for Sjögren-Larsson syndrome. He joined the University of Nebraska Medical Center in 2002 as Professor of Pediatrics and assumed Directorship of the Metabolic Diseases Clinic at Children's Hospital. In this capacity, he oversees the care of children with a variety of inherited metabolic diseases and performs laboratory research in the area of biochemical and molecular genetics.

What is Cystinosis? Cystinosis is a rare, inherited, metabolic disease that is characterized by the abnormal accumulation of the amino acid cystine in each cell. The build-up of cystine in the cells slowly and 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 less than 2,000, worldwide. It is one of the 6,000 rare or "orphan" diseases in the United States that collectively affects more than 23 million Americans.

2006 RESEARCH STUDIES FUNDED AND COMMITTED: \$2,313,962

FEBRUARY 2006

Ranjan Dohil, MD

University of California, San Diego

“A Study to Evaluate Enteric-Coated

Cysteamine Therapy in Patients with Cystinosis”

\$253,685 – 1-year clinical study

MAY 2006

Corinne Antignac, MD, PhD

Hospital Necker, Paris, France

“Characterization of Cystinosin Intracellular Trafficking”

\$85,000 – 1-year study

Angela Ballantyne, PhD and Amy Spilkin, PhD

University of California, San Diego

“Academic Functioning in Cystinosis:

A Comprehensive Study of the Process of Achievement”

\$213,527 – 2-year study

Bruce Barshop, MD, PhD and Jerry Schneider, MD

University of California, San Diego

“API-4000 Tandem Mass Spectrometer for Cystinosis Research”

\$118,400 – 2-year lease and maintenance

Elena Levchenko, MD, PhD

University Medical Centre St. Radboud Nijmegen, The Netherlands

“Pathogenesis of Interstitial Renal Damage Leading

to Renal Failure in Cystinosis”

\$72,423 – 1-year study

Daniel Salomon, MD and Stephanie Cherqui, PhD

The Scripps Research Institute, La Jolla, California

“Treatment of Cystinosis Nephropathy Using Genetically

Modified Adult Stem Cells in Murine Cystinosis Model”

\$709,170 – 3-year study

DECEMBER 2006

Rita Ceponiene, MD, PhD

University of California, San Diego

“Neural Functioning in Auditory and Visual Systems

in Cystinosis: Linking Brain to Behavior”

\$287,918 – 2-year study

Francesco Emma, MD, Anna Taranta, PhD and Elena Levchenko, MD, PhD

Bambino Gesù Children’s Hospital and Research Institute, Rome, Italy
and Radboud University Medical Centre Nijmegen, The Netherlands

“Transcriptional and Post-Transcriptional Regulation of the CTNS Gene”

\$138,824 – 2-year study

Eric Moses, PhD, Mentor

Katy Freed, PhD, Research Fellow

Southwest Foundation for Biomedical Research, San Antonio, Texas

“Complex Genetic Approaches to Monogenic Disease: Genomic and

Transcriptomic Dissection of Normal Expression of CTNS, the Gene

Involved in Nephropathic Cystinosis”

\$148,535 – 2-year study

Vasiliki Kalatzis, PhD, Mentor

Claire Hippert, Pre-doctoral Research Fellow

Institut Génétique Moléculaire Montpellier, Montpellier, France

“Gene Transfer Studies for Cystinosis”

\$168,290 – 2-year study

Holger Willenbring, MD, Mentor

Kentaro Okamura, PhD, Research Fellow

University of California, San Francisco

“Targeted Cell Fusion for the Correction of Tubulopathy Due to Cystinosis”

\$84,530 – 2-year study

Jess G. Thoene, MD

The University of Michigan

“Lysosomal Cystine Enhanced Apoptosis

in Cultured Human Mesenchymal Stem Cells”

\$33,660 – Laboratory supplies

*Jess G. Thoene, MD

(Reinstated after Hurricane Katrina)

The University of Michigan

“Lysosomal Cystine Enhanced Apoptosis

in Cultured Human Mesenchymal Stem Cells”

\$150,000 – 18-month study

** Dr. Thoene’s study was originally approved for funding in 2005 but because his lab at Tulane University was destroyed by Hurricane Katrina the study was cancelled.*

In November 2006, Dr. Thoene’s study was reinstated at the University of Michigan.

To review the full abstract on any of the studies see the *Cystinosis Research Foundation Science Report* or visit www.natalieswish.org.

Cystinosis Research Foundation Fellowship Program

In 2006 the Cystinosis Research Foundation established the first Cystinosis 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. Applications will be available in conjunction with the spring and autumn Call for Funding Proposals.

2007 Call for Funding Proposals

The Cystinosis Research Foundation will announce a Call for Research and Fellowship Proposals in the spring and autumn of 2007. The CRF is prepared to fund proposals to improve the immediate care of children and young adults with Cystinosis and to develop new understanding and treatment of Cystinosis to help these children. The number of awards and their value will depend on the number of outstanding proposals received and the funds available in 2007.

The Cystinosis Research Foundation is currently planning the first International Cystinosis Research Foundation Symposium scheduled for spring of 2008.

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My Family's Story

KEVIN, TERESA, PATRICK AND JENNA PARTINGTON

Hi! My name is Jenna McCall Partington and I am two years old. My mom wanted me to let you know a little bit about our family and I am happy to tell you our story.



I was born on December 7, 2004, one minute after my brother Patrick Douglas. At 6 lbs. 10 oz. and 5 lbs. 9 oz., we were pretty big for twins. We did just great our first few months of life. However, when we turned 6 months old, we stopped growing. Also, my brother was just a geysier, throwing up all the time! Mom let him taste water drizzled out of a straw at a pizza place one day, and he went crazy. He sure seemed thirsty. We both started going to the doctor an awful lot. Something just wasn't right. Last March I was hospitalized with Rotavirus for 12 days. I became so dehydrated that I was moved to ICU. It was a horrible time for all of us...but because of that sickness and all of the good doctors, we finally figured out that I had Cystinosis. My brother's diagnosis was confirmed as well.

My parents moped about the house after learning of our rare and incurable sickness. Mom read Norman Vincent Peale's *The Power of Positive Thinking* and hopes to make its teachings our family's way of approaching life with Cystinosis. We are only two, and we never know what the future may bring. However, Mom and Dad tell us to be strong and grateful for all we have. We take our 10 daily medicines very well. I love water and my brother and I love nothing more than a cool new cup or glass. I overheard Mom and Dad talking and they said the hardest thing right now is keeping up with our demand for water and our 12 or so diaper changes each per day. What will they do with themselves when we can get our own water at the sink and use the potty?

My Mom and Dad have some really special friends, who created **Jenna & Patrick's Foundation of Hope** following our diagnosis. Successful fundraisers have been *Chip in Fore Charity Event* and a *Walk of Hope!* My parents and their friends are excited to pass these funds along to the Cystinosis Research Foundation this year at the Natalie's Wish event on June 1 in Newport Beach. Patrick and I would like to come, too, and meet some other kids with Cystinosis, and meet the very special doctors who are working so hard to make us better.

I, personally, look forward to meeting Natalie. I have only heard of her from Mom and Dad, and she sounds like such a special person. She is brave (and so pretty!) and does so much to make a difference for all of us who have Cystinosis. There are a lot of other kids, families and doctors out there who are working so hard and giving so much of themselves to this disease. I have nothing but HOPE that things are going to be all right!

Mom and Dad want me to say: "Thanks to Nancy, Jeff, Alex and Natalie for your efforts at creating awareness, bringing doctors together and producing progress to the world of Cystinosis."

Lots of love to our "Cystinosis Family" out there.

Jenna

This year we are launching a new fundraising event—our inaugural Fore A Cure Natalie's Wish Golf Tournament to help children and young adults diagnosed with Cystinosis. The tournament will be held Monday, October 1, 2007 at the exclusive Coto de Caza Golf & Racquet Club.

Our goal is to provide a golf tournament experience for Orange County's top business and community leaders, who will enjoy a first-class golf course, camaraderie, prizes and an opportunity to make a wish come true for those who suffer from Cystinosis. The entry fee of \$2,500 includes golf for four, gift bags, putting and on-the-course contests, hole-in-one contest, lunch, cocktail reception, silent and live auction, dinner and awards.

To join our committee, volunteer at the event or become a sponsor, contact Zoe Solsby at 949.223.7610 or visit www.natalieswish.org.

Families Unite to Find a Cure for Cystinosis

Just last year, Teresa and Kevin Partington's twin 13-month-old children, Jenna and Patrick, were diagnosed with Cystinosis. The Partington's family and friends quickly rallied around them in support and formed the **Jenna & Patrick Foundation of Hope (JPFH)** to raise money for Cystinosis research.

The foundation has been very successful and has recently announced that the majority of funds donated to JPFH will be donated to the Cystinosis Research Foundation in an effort to streamline and expedite Cystinosis research around the world. One hundred percent of the money received from the JPFH will go directly to Cystinosis research and the Cystinosis Research Foundation Fellowship Program.

The Cystinosis Research Foundation and Jenna & Patrick Foundation of Hope are intricately united by the race to save our children's lives and our quest to find better treatments and a cure for all those who suffer from Cystinosis. We are honored to collaborate with the Partington family and their foundation.



Monday, October 1, 2007