

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 ★ SPRING 2005



From Natalie

- ❖ *I started school in September and am now in the eighth grade. My favorite subject in school is French. I am*
- ❖ *enjoying school with all of my friends and teachers.*

In October I volunteered for Dr. Dohil's study. He hopes to find a time-release medication for Cystinosis patients. I went to the hospital and stayed for six nights. At first, I hesitated to volunteer because I was afraid and did not want to be in any pain. I decided to volunteer because I knew it would help many people with Cystinosis and people with other diseases. It can help benefit the world.

The first day, Dr. Dohil inserted the end of a six-meter tube down my nose. I also had an I.V. so they could draw blood easily. Each day they pushed the tube down further until all of the tube was in my stomach and intestines. I only felt a little pain. After they pushed it down, they would give me my medicine through the tube and then draw my blood. They took more than 120 vials of blood, but I am used to blood draws so it did not really bother me. The last day they had to take the tube out of me. It tasted like chicken, Godiva chocolate and moldy pasta when it came out! It only hurt a little.

I am glad I did the study because I know that it will help find a time-release medication. If the doctors can make a time-release medication, I will not have to take medicine every 6 hours, every day. I will only need to take it twice a day, which would be a lot easier, especially at school.

Thank you all so much for giving all the donations because it will really help find a cure and a time-release medication. I always pray for a cure. I also thank God for letting me be so healthy and so fortunate to have so many people who support me with their generous gifts. Love, Natalie



Natalie and friends at her 14th birthday party

Dear Friends and Family

What a year it has been! Our annual Natalie's Wish event in May of 2004 was an enormous success. The event, held at the Pelican Hill Clubhouse, was attended by over 250 people. Our guest speaker was Dr. William Gahl, Clinical Director of the National Human Genome Research Institute and the Director of the Intramural Office of Rare Diseases at the National Institutes of Health (NIH) in Bethesda, Maryland.

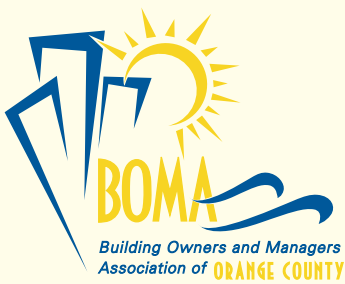
Dr. Gahl spoke to the guests about rare diseases and the need for research to study and understand such diseases, which collectively affect more than 25 million Americans. Dr. Gahl explained what Cystinosis is and shared a slide show that illustrated a variety of rare diseases. Dr. Gahl has been involved in Cystinosis research for over 23 years. He continues to see Cystinosis patients at the NIH.

The evening culminated with a Fund-A-Cure round of donations totaling an amazing \$243,000. This year as in years past, Mike and Lynette Hayde made a generous matching gift challenge. Their \$50,000 challenge was doubled by John Hagestad and Bill Thormahlen of SARES-REGIS Group, who increased the matching gift by an additional \$50,000. The \$100,000 challenge was quickly met and by the end of the evening we had raised over \$243,000 for Cystinosis research! We cannot thank you enough for your support in our efforts to find better treatments, and ultimately a cure for Cystinosis.

In response to our Thanksgiving newsletter we received donations of over \$32,000, which brought our year-end total to an incredible \$695,000. What a difference you have

made to further our mission and goals. Since 2003, the Cystinosis Research Foundation has funded four important multi-year research studies.

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BOMA Scores for Natalie

The Building Owners and Managers Association (BOMA) of Orange County will be hosting the BOMA Annual Charity Golf Tournament and Auction on August 2, 2005 at Coyote Hills Golf Club in Fullerton.

The golf committee and Board of Directors have again whole-heartedly embraced the Cystinosis Research Foundation as the beneficiary for all funds raised at the live and silent auctions. In 2004, BOMA-OC raised over \$12,000 for Cystinosis research!

We are grateful for the tremendous support of all who participated in last year's event. If you would like to play in this year's tournament or provide an auction item, contact Mike Raring at AAA Electric & Communications at 949.279.0594 or email him at mraring@aaaelectric.com.

With your assistance we will make the 2005 Charity Golf Tournament the most successful yet bringing Natalie's Wish closer to reality.



Retired Marine Corps Sgt. Major Jim Morris in a pre-race photo—wearing a picture of Natalie—with his daughters Denise (l) and Deborah (r) as they prepare to brave the weather in Huntington Beach.

Braving the Rain for Natalie

Retired Marine Corps Sgt. Major Jim Morris, along with his beautiful daughters, Denise and Deborah, braved the cold winter rain and strong

winds on Sunday, February 6 in Huntington Beach to run a half-marathon on behalf of Natalie and the Cystinosis Research Foundation.

The Pacific Shoreline Marathon drew more than 7,500 participants, with the Sgt. Major coming in fifth for his age division. His wife Betty was the team coach providing water, food, a change of clothing and blankets.

The day started at 0300 wake up and on the road by 0345. The race began at 0700 and 1 hour 22 minutes and 40 seconds later the Sgt. Major crossed the finish line duplicating his personal best time from October of last year. Deborah ran a 5K and Denise ran the 13K—both women finished in good time under difficult conditions!

We are grateful to our friends, the Morris Family, for their efforts to help Natalie and others with Cystinosis.



Last spring, after successfully selling cookies for the first time, the twenty-three Brownies of Troop #163 of Harbor Day School gathered in small groups to discuss how they might distribute some of their proceeds to help others.

Brownie Katie Flattum, shared that she had read the Cystinosis Research Foundation newsletter and thought that it would be a good idea to help the foundation since Natalie Stack was a fellow Harbor Day student. All the girls agreed and were pleased to send a donation to the foundation.

The girls also received a meaningful gift in return—a kind letter from

Natalie thanking them for their contribution.

The letter is now a part of each girl's Brownie scrapbook.

Cookies and Brownies for Natalie

Rebecca Beyer and Samantha Mangione



Two Teens Give Out Party Favors

Rebecca Beyer and Samantha Mangione, two friends who shared their 14th birthday party at the Coastal Canyon Clubhouse in Newport Beach, celebrated by sharing the best gift of all—the gift of giving. Each of the girls chose a charity to receive the donations in lieu of gifts. Rebecca, who attends Harbor Day School, and has gone to school with her friend Natalie since kindergarten, chose the Cystinosis Research Foundation. “The best gift I can have is to bring the cure to Cystinosis a little closer,” said Rebecca. Samantha chose Ronald McDonald House. The girls were recognized in the *Daily Pilot* for their party about giving, not receiving.



We are proud to report that 100 percent of your donations are used to support medical research. With your commitment to our vision and our goal we are closer to realizing Natalie’s Wish.

Thank you for your support.

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Spring Clean-Up Weekend

In the Newport Beach neighborhood of Harbor View Homes, the first weekend in April is designated as the annual Spring Clean-Up Weekend. Hundreds of families participate in this event.

Last spring, two very thoughtful children, Allison and Austin Ridge, ages 10 and 7, did a remarkable thing. They set up a stand in front of their house, and sold dog leashes and dog beds to the crowds that shopped the streets. The items were provided by the children’s uncle who owns an animal product business. A portion of the proceeds were generously donated to The Cystinosis Research Foundation. The children raised more than \$200, which exceeded their expectations.

Allison and Austin got the idea after seeing the *Natalie’s Wish* newsletter that featured Natalie Stack, who is a cousin of the children’s good friends Maddie and John O’Connor. Both children wanted to know more about the disease and wondered how they could help Natalie. Shelley and Steve Ridge are very proud of their children and remarked that it was “fun to see them so excited about being able to help Natalie and other children with Cystinosis.”



Donor list continued on next page



Senator Kennedy with Natalie and Alex Stack

Did You Know...

The Stack Family was honored to meet Senator Edward Kennedy on his recent visit to California. They thanked him for his commitment to the 25 million Americans who suffer from rare diseases. In 2001, Senator Kennedy and Senator Orrin Hatch introduced The Rare Disease Act (RDA) of 2001, which enhances the prospects of developing new treatments and cures for the over 6,000 rare diseases in the United States.

The RDA provided authorization for the Office of Rare Diseases (ORD) at the National Institutes of Health. It authorized regional centers of excellence for rare disease research and increased funding for ORD. The RDA provides vital research for rare diseases and disorders.

Cystinosis afflicts approximately 500 people, mostly children in North America, and less than 2,000 worldwide. It is a rare genetic disorder that causes cystine to accumulate in organs. Cystine is trapped in the cell and forms crystals causing cell death as its concentration increases. The crystals slowly destroy organs in the body including the kidneys, liver, eyes, muscles, thyroid, white blood cells and the brain.

Daily life is painful for children with Cystinosis, and unfortunately, there is no cure. Cystinosis affects so few people that research money is scarce and progress is slow. But there is hope.

Insights gained from research on diseases such as Cystinosis help to understand other diseases. Cystamine is being considered as a treatment for Huntington's disease. In a mouse line genetically manipulated to serve as a model of Huntington's disease, cystamine improves neurological symptoms and prolongs life. Cystamine is the oxidized form of cysteamine, which was developed for Cystinosis. Continued research will determine if this compound will help those with Huntington's disease.

For more information about "orphan" diseases: visit www.rarediseases.net.

Donor List *continued from previous page*

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We deeply appreciate your support.

Special thanks to all those who have volunteered their time and talents for Natalie's Wish. A special thank you to Kris Elftmann of Noelle Marketing Group for his creative talent, vision and personal dedication to the Cystinosis Research Foundation.

The new web site design was generously donated by Amies Communications. We are grateful to Grif Amies, Randy Taylor and Rob Gonzales for their commitment to CRE.

We have attempted to include every donor on this list. We apologize for any errors or omissions. Please contact **Zoe Solsby** at **949.809.2400** if we have listed you or your donation incorrectly.

Friends Updates



SHANNON PAJU

Many of you met Shannon Paju at last year's Natalie's Wish event. Shannon spent many weeks in the hospital during 2004, but she was determined to graduate from high school in June – which she did! Congratulations Shannon! In her own words...

“After last year's event, I went back in the hospital for eighteen days in June. I was out for one week and returned to the hospital for another five days. I was in the hospital on June 14, but I was able to attend my graduation where I sang a solo. I returned to the hospital after the ceremony.

Throughout the year, I was hospitalized on several occasions but I was able to spend some time with my friends. I also attended two classes at Cypress College.

My transplanted kidney isn't doing well and I am back on the transplant list. I hope and pray that 2005 will be a better year for me.”

ZACQARY BEERS

Zacqary has been doing very well. Zacqary is in his second year of pre-school, and his teacher and therapists love him. Although he is rambunctious at home, he is obedient at school, and loves the structure. He rides the bus to and from school, which is his favorite part of the day.

He is now in the 75th percentile for height because of the growth hormone injections he receives daily. He enjoys riding his bike and running around the park with his sister, Zoe, but his true love is movies.

Zacqary loves animals. He visits his grandparents' dog at least once a week. He will be getting his own dog soon because the interaction brings out his loving nature.



Scientific Review

Board Collaboration Announced

We are pleased to announce that in October, 2004, the Cystinosis Research Foundation (CRF) and the Cystinosis Research Network (CRN) announced plans to jointly utilize the established CRN Scientific Review Board for all research proposal reviews. CRN is a volunteer, non-profit organization headquartered in Burlington, Massachusetts. The CRN supports Cystinosis research; provides family support and assistance; and is active in community awareness and education programs.

The Scientific Review Board is comprised of leading Cystinosis researchers, scientists and doctors. The Board will review all grant proposals; advise the Cystinosis Research Foundation on the scientific merit of each proposal; submit funding recommendations; and offer general research guidelines.

The Scientific Review Board provides independent, objective review and recommendations regarding each proposal submitted utilizing grant review guidelines established by the CRN and CRF. With all research proposals now being reviewed by a single Scientific Review Board, the process will result in more efficient funding. Both the Cystinosis Research Foundation and Cystinosis Research Network are confident that the collaboration will accelerate all areas of Cystinosis research.

Visit www.natalieswish.org for more information about the research guidelines.

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Cystinosis Research Foundation

Funded Studies—Update

For a full review of our funded research visit www.natalieswish.org

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and Hepatology, UCSD School of Medicine*

Dr. Dohil was awarded a two year grant by the Cystinosis Research Foundation. The purpose of his study was to determine where cysteamine was absorbed within the intestinal tract. His long term aim is to identify an alternative way of delivering cysteamine so that it is as effective or more effective than the current regimens and can be administered less often through the day, thus eliminating the unwanted side effects such as bad odor and gastrointestinal symptoms.

The study is going very well. Dr. Dohil and his team are close to completion and have so far enrolled nine adult control subjects and six children with Cystinosis. Each subject is asked to stay in the hospital for six to eight days during which time a specially designed tube, passed through the nose, is allowed to pass all the way down to the large bowel. At different points during its descent, cysteamine is injected into the tube and then measured by serial blood samples to see how well cysteamine is absorbed.

The preliminary data demonstrates that the best absorption of cysteamine is in the small intestine, particularly within the lower small intestine. Dr. Dohil is still analyzing data regarding the efficacy of cysteamine when it is administered at different sites in the intestinal tract. The data at this point suggests that a sustained-release or enteric-coated dosage form of cysteamine is feasible. There still, however, remains a considerable amount of work to be done. Once the data is fully analyzed Dr. Dohil plans to talk with appropriate pharmaceutical companies to discuss future options.

Doris A. Trauner, MD

*Professor, Departments of Neurosciences
and Pediatrics; Chief, Pediatric Neurology
UCSD School of Medicine*

Dr. Trauner was awarded a two year grant by the Cystinosis Research Foundation to study myopathy (muscle wasting). The purpose of the study was to specifically determine whether there is evidence of mitochondrial dysfunction in Cystinosis myopathy and to study the potential beneficial effect of a “mitochondrial cocktail” containing Coenzyme Q10 and multiple B vitamins as well as L-carnitine, on muscle function. Dr. Trauner is hoping to understand why muscle wasting occurs and determine a

treatment regimen that will improve strength and prevent deterioration in muscle strength.

To date, ten patients have been enrolled in the study and five patients have completed the first data point of testing. Currently, four of the patients have been started on the randomized experimental therapy or placebo, and will be returning for their second data point within the next 3-4 months. A second group of five patients are scheduled to be in San Diego for baseline studies within the next two months.

Preliminary results show that three of the five subjects studied to date have had evidence of a myopathy on both EMG and muscle histopathology. The EMG on two of the patients also demonstrated neuropathic as well as myopathic changes. If these findings and other preliminary findings are consistent across the larger group of adults with Cystinosis to be studied, they will provide valuable new information regarding the cause of the myopathy, as well as whether treatment with a “mitochondrial cocktail” will have a positive effect in reducing the symptoms of the myopathy.

Thomas Jeitner, PhD

*Research Assistant Professor, Department of
Biochemistry, Medical College of Wisconsin*

Dr. Jeitner was awarded a two year grant by the Cystinosis Research Foundation. He has dedicated the majority of his career on the biochemical pharmacology of cysteamine and its analogs. The purpose of his study is to discover better drugs for the treatment of Cystinosis.

In particular, Dr. Jeitner is focusing on the delivery method of cysteamine (Cystagon™) by cystamine. Dr. Jeitner’s study focuses on three aspects of the actions of cysteamine and its metabolism. The first is to understand why animals can tolerate greater amounts of cystamine than cysteamine. The end result is that cystamine acts as a slow-release delivery system. The second is to study cell death. Preliminary results show that cystamine prevents the form of cell death that is more likely to be acting in the kidneys of patients with Cystinosis. This is a very important finding because it has provided a better idea of cystamine’s unique effects

on the body and has suggested some alternate treatment strategies for Cystinosis. The third aspect of the study has led to a discovery that is the most exciting of all. Dr. Jeitner and his team have discovered that cystamine generates a gas that can convert the damaging cystine into cysteine. The gas in question can be delivered by a number of other agents that might provide better treatments for Cystinosis.

Dr. Jeitner’s studies are restricted to test tube studies and cell culture experiments. Test tube studies ensure that the chemistry is right. The cell culture experiments ensure that the biology is sound. The next phase is to test the above ideas in animals and this is what Dr. Jeitner is preparing to do. Dr. Jeitner’s studies will be the basis of an application to the National Institutes of Health later this year to further fund Cystinosis research.

Jess G. Thoene, MD

*Director, Hayward Genetics Center
Karen Gore Professor of Pediatrics
Tulane University, School of Medicine*

Dr. Thoene has been awarded a one year grant by the Cystinosis Research Foundation to study how lysosomal cystine causes enhanced apoptosis (early programmed cell death). When Dr. Thoene studied apoptosis in cultured Cystinosis skin cells, he found that the presence of lysosomal cystine increases the proportion of cells entering apoptosis by two hundred percent. Over time, this inappropriate commitment to cell death can reduce the number of remaining cells leading to generalized hypocellularity, and the short stature, failure to thrive and most importantly, the kidney disease seen in nephropathic Cystinosis.

This year, Dr. Thoene has begun the process of inquiring into the actual molecular mechanism whereby lysosomal cystine augments apoptosis. Apoptosis is a complex process with more than 300 proteins involved. The long term goal of this project is to understand how lysosomal cystine storage in Cystinosis causes the lethal nephropathic phenotype. Dr. Thoene’s immediate goal is to elucidate the mechanism(s) whereby lysosomal cystine storage markedly increases the rate of apoptosis in cultured cells.



Photo by Sonal Vyas

Jeff and Nancy Stack with Dr. Ranjan Dobil (left) and Dr. William Gabl (right) at the Natalie's Wish 2004 event

Dear Friends *continued from front page*

The Cystinosis Research Foundation is the largest nonprofit fund provider of Cystinosis research. As of December 2004, the Foundation has funded and committed more than \$736,000 supporting several Cystinosis research projects. This year we have more than \$420,000 available for new research projects. We recently announced to the medical and scientific community that we are accepting grant proposals and fellowship applications. On June 2, during our annual Natalie's Wish event, we will announce which research studies will be funded in 2005.

We are fortunate to have so many dedicated scientists and doctors working on better treatments and a cure for Cystinosis. We were thrilled to attend the first symposium on Cystinosis in May, 2004. The conference was held at the National Institutes of Health in Maryland. It was sponsored by the National Institutes of Health and Office of Rare Diseases. The conference was attended by doctors and scientists from around the world. Parent groups also attended and were able to present first-hand knowledge of what it is like to live with Cystinosis on a day-to-day-basis.

The conference provided an opportunity to share information, discuss ideas, and collaborate on Cystinosis research and treatments. It was an incredible opportunity for the Cystinosis Research Foundation to be part of such a prestigious, committed group of individuals who are dedicated to improving the quality of life and prolonging the lives of our children.

To those who attended the 2004 Natalie's Wish event and to the many others who have made contributions, we thank you for your generosity. We could not do this without all of you. We continue to be amazed by the outpouring of love, concern and support. You have allowed us to share this journey with you and in doing so, have given us strength and support. We are so very blessed to have such a wonderful community of family and friends.

Your financial support, good wishes and prayers have provided Natalie and others with Cystinosis the hope and support they so desperately need. We thank you for your commitment to our cause. We are also grateful to the many volunteers who made our Natalie's Wish event such a success. We are especially thankful for the tireless efforts of Zoe Solsby, Marylyn Milburn and Vince Ciavarella.

We hope you will join us for this year's Natalie's Wish event on Thursday, June 2, 2005 at the Four Seasons Hotel, Newport Beach. The evening will begin at 5:30 with wine and hors d'oeuvres followed by research updates, the new Cystinosis video and Fund-A-Cure. We look forward to seeing you there.

Natalie on the Net

We are excited to announce the launching of our new Natalie's Wish web site. Our goal is to provide information about Natalie, Cystinosis, our funded research, upcoming events and news. We have designed the site for easy navigation and will be updating it frequently. Please visit the site www.natalieswish.org and fill out the contact information form and provide us with your comments. We look forward to hearing from you!

I wish I may, I wish I might, have the wish I wish tonight.

CYSTINOSIS RESEARCH FOUNDATION

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THE FOURTH ANNUAL
"NATALIE'S WISH" FUNDRAISER
THURSDAY, JUNE 2, 2005
FOUR SEASONS HOTEL
NEWPORT BEACH, CALIFORNIA

FOR INFORMATION ABOUT THE EVENT
VISIT WWW.NATALIESWISH.ORG