Happy 10th Birthday,
Cystinosis Research Network!

By Mary Beth Krummenacker

When I began to think about writing the story of the last 10 years of the Cystinosis Research Network, I suddenly felt overwhelmed with so many different thoughts and emotions. TEN YEARS – how could that be? Where did this all begin? What have we done? Who have we helped? Where are we going? Why do we do what we do?  And then I realized why—my daughter Laura is why. Laura is a 20 year old college student who works two jobs and enjoys life. She also happens to have cystinosis, and if it wasn’t for this organization and the love and support of the incredible people involved -Laura and I wouldn’t be where we are today!

Cystinosis Research Network (CRN) began with a vision. The vision of two people – Jack & Colleen Hammond. What began as a common bond between the Hammonds and I – children with cystinosis – soon grew into a friendship. As time went on, we became closer friends. I would often travel to the Boston area to support them in their fundraising efforts. I would help in any way I could and was always so appreciative of the efforts of their friends, family, and community in their response to the cause of cystinosis. As our network of families grew, we began to see the need to do more. We needed to be much more pro-active and expect more of our scientific community and in our own way, demand more be done. Cystinosis had a treatment, but it didn’t have a cure. We began to grow impatient and noticed that the interest in cystinosis research was waning. Instead of looking for that elusive cure, people were becoming complacent. We were concerned for the future for our children and realized that if we didn’t step up to the plate and set that agenda, no one else was going to. With the guidance and advice of some of our scientific community, we formalized ourselves into Cystinosis Research Network and began setting forth with an agenda that has been filled with many wonderful accomplishments.
Message from the President
José T. Morales

I trust that all of you are fully engaged with life's challenges...new academic years, birthday celebrations...and of course the on-going management of your own or your children's medical needs. If you are anything like us, we continue our quest to self-educate ourselves on how best to meet the needs of our children and to maintain a sense of "normalcy". We rely on an extensive network of resources to ensure we are prepared and informed when we have to make decisions regarding our child's health.

The Cystinosis Research Network is a primary source we rely on. The Cystinosis Research Network’s mission is comprehensive. We focus on meeting all of our community's broad needs. We are your advocate to the medical community, we are your advocate to the pharmaceutical industry, we are your advocate to the government, we are...

Vision – The Cystinosis Research Network vision is the discovery of improved treatments and ultimately a cure for cystinosis.

Mission - The Cystinosis Research Network is a volunteer, non-profit organization dedicated to supporting and advocating research, providing family assistance and educating the public and medical communities about cystinosis.

Research – In 2006, we have funded $195,000 of research. Today CRN is sponsoring seven individual research projects. This translates to a financial commitment of $472,000 during the next four years.

Research Studies:

- Dr. Trauner - “Early Intervention Trial for Visual Processing Deficit in Cystinosis”
- Dr. Thoene - “Increased Apoptosis Produces the Phenotype in Nephropathic Cystinosis” (Study is Complete)
- Dr. Thoene - “Tissue Repository for Cystinosis”
- Dr. Antignac - “Development and in vivo Testing of Novel Therapies for Cystinosis”
- Dr. Cairns - “Design and Synthesis of Novel Prodrugs for the Treatment of Cystinosis”
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- Dr. Trauner - “Cognitive Profiles in an International Sample of Individuals with Cystinosis” (Study is Complete)
- Dr. Levchenko - “Study of ATP metabolism in human cystinotic proximal tubular cells and in humans with cystinosis in vivo”
- Mr. Nischal - “Development of a Cysteamine in situ gelling system for the topical treatment of corneal crystals in Cystinosis”

We are excited about the ever expanding research efforts and continuing progress being made.
Development – John Shepperd took on the responsibilities for this committee in the beginning of the year and is in the process of elevating the processes and protocols via which we raise the necessary funds to meet all of demands of the organization. In 2006, we set two objectives: a target of $250,000 and a stretch target of $300,000. We have raised 90% of the target funds to date. We have two major fundraising events on the calendar and believe we will achieve the target.

Education and Awareness – We continue to place a high degree of emphasis on making the medical professionals aware of cystinosis. The critical networking and outreach CRN continues to do at these conferences is invaluable. It is hoped that we can maintain this high level of visibility for 2007. We participated or maintained a booth at all of these targeted conferences and attended an International Family Conference in 2006:

University of Miami Pediatric Nephrology (March 9 – 14), Miami Beach, Florida
American Society of Pediatric Nephrology and Pediatric Academic Societies (April 30 – May 2)
San Francisco, California
Netherlands Cystinosis Foundation Family Conference (June 30 – July 2)
Genetic Alliance Annual Conference (July 27-29) Bethesda, Maryland
American Society of Nephrology, (November 16-19), San Diego California

Family Support - We have had a lot of success in providing support to a number of individuals from our community. Support has come in the form of fiscal and moral support.

2007 Searching for A Cure in the Lone Star State Medical and Family Conference – The date has been set. The city and venue have been selected: July 19-21, 2007, The Hotel Contessa, San Antonio, TX. We are in the midst of planning the agenda and the necessary logistics. Your 2007 Family Conference Committees:

Host Family  **John and Kim Shepperd**
Co-Chairs  **Christy Greeley, Marybeth Krummenacker and Pam Woodward**
Agenda  **Christy Greeley, Marybeth Krummenacker, José Morales, and Pam Woodward**
Day Care  **Tonya Wise, Lynn Thomas and Sandy Glaize**
Marketing  **Pam Woodward, Marybeth Krummenacker, Kathleen Harrison, Denise Flechinger, and Carol Hughes**
Speaker/ Advisory Board  **Christy Greeley, Elva Smith, and Marybeth Krummenacker**
Sponsorship  **Mack Maxwell, Dave Greeley, and José Morales**
Welcome/ Registration  **Frankie McGinnis, Cheri Friend, Melissa Morgan, Karen Gledhill, Paula Shal, and Lorna Smith**
Site Planning  **John and Kim Shepperd, Pam Woodward, and Dan Julian**

**Note: Bold and italics denote leads**

The committees have been formed and are making good progress. We will be communicating more specifics as the plans become more firm. We hope to have a strong attendance, so start polishing those cowboy boots and dusting off those ten gallon hats!

It takes a lot of people working together to make all of this happen. It is true our numbers continue to grow, but we also have greater and greater needs. There exists a multitude of ways to participate in the search for a cure for cystinosis, and none are too small. We respectfully ask that all of you seek a way to become engaged and contribute.

José T. Morales
President
Happy 10th Birthday, CRN!

Over the last 10 years, we have been the beneficiaries of some fantastic fundraisers, from Garage Sales to Dinner Dances, from a Hoe Down to a High School Spirit Week, from a Family Fun Day to Golf Outings and so many more incredible fundraisers. Each and every one of these fundraisers has helped CRN over these last 10 years in delivering our message of new and improved treatments and have been so important to the growth of CRN. So many wonderfully, generous, and kind people have touched the lives of families they don’t even know. Most importantly we have supported research and researchers throughout the world with those funds raised. We have been able to do so many other remarkable things as a direct result of those events, from establishing an online support group, to a twice a year newsletter, to exhibiting at medical conferences. Whether it is the American Society of Pediatric Nephrology or the American Academy of Pediatrics, at these medical conferences we have shared thousands of copies of our brochures, our newsletters, important current cystinosis research articles, and we have had the experience of networking with doctors from around the world to personally share the cystinosis story. We have networked with the pharmaceutical industry – in particular Sigma Tau Pharmaceuticals – who have been extremely generous with their support financially to CRN.

In addition, one of the most important things we have done was to encourage the FIRST ever Scientific Symposium held at the NIH. That meeting was an unprecedented event that brought together over 30 of the world’s leaders in cystinosis research to discuss the past, present and future of cystinosis. CRN was represented at that meeting by two board members and hosted a wonderful dinner where these amazing people could sit and informally discuss their work and ideas. CRN has also established an active Scientific Review Board who continually look at proposals for new research and always with the CRN mission statement at hand - “looking for new and improved treatments and an eventual cure” to this devastating disease.

CRN has hosted two major family conferences over these 10 years – one in Florida and one in Utah - with the third one being planned for Texas, as we go to press with this newsletter. Each of these conferences have built on the other and have turned into incredible events for our families to come together – some for the first time – to meet and network with others who truly know what it is like to live with this disease called cystinosis.

As we look to the future of cystinosis, I feel more hopeful than ever before! This extraordinarily small organization has done extraordinary things in its very short 10 years ----- let us continue with that vision and work together to give our loved ones the best and the brightest future we possibly can. In the words of Margaret Mead – “Never doubt that a small group of thoughtful, committed citizens can change the world. Indeed, it is the only thing that ever has.” Let us – together – continue to change the world of cystinosis!
The Cystinosis Research Network is pleased to announce its Medical and Family Conference to be held at the Hotel Contessa in San Antonio, Texas from July 19 - July 21, 2007. People from all over the world will come together for three days of exciting lectures, workshops and sharing. Clinicians and researchers will present leading edge information, answer questions and share their insights. All of this will take place in beautiful San Antonio, Texas at the Hotel Contessa, located on the Riverwalk in the heart of San Antonio (www.thehotelcontessa.com). A reflection of the state's most popular destination, this contemporary 12-story property captures both the historic and cultural essence of San Antonio. From the atrium lobby overlooking the Riverwalk, and the warmth created by the Mediterranean design, to the overall comfortable elegance, Hotel Contessa establishes a friendly, welcoming environment from the moment of your arrival.

Please join us for what promises to be a tremendous conference filled with new understanding, new awareness, and new friends. You will have the opportunity to meet and network with people who empathize with your experiences, and learn from and speak with the world's top cystinosis clinicians and researchers. More specific registration and agenda information will be mailed to families and posted on the CRN website (www.cystinosis.org) in the near future.
Meet the New CRN Board Members

Christy Greeley - Executive Director

Christy Greeley, who has served on the Cystinosis Research Network Board of Directors as Vice President of Research since 2003, has accepted the additional position of Executive Director. As Executive Director, Christy will act as the chief administrator of CRN and will be primarily responsible for the overall operation of CRN and supervision of its staff and programs.

Christy graduated from the University of Michigan with a Bachelor of Science in Psychology and a Master of Public Health in Public Health Policy and Administration. She has worked in both laboratory and clinical research at the University of Michigan Medical Center in the departments of Rheumatology, Pathology, and Psychiatry. She also worked for 9 years in pharmaceutical research at Parke-Davis Pharmaceutical Research as a Clinical Research Associate and Clinical Scientist, managing clinical trials for a variety of central nervous system drugs.

Christy's husband, Dave, is also a CRN Board Member. They have a son, Jack, and a daughter, Alex.

Brittney LeBeau - Treasurer

Brittney LeBeau is from Aurora, Illinois and married to her high school sweetheart, Brett. The LeBeaus have two children, Alexis and Evan, ages 5 and 3 respectively. Both have Cystinosis and were diagnosed almost 3 years ago. They are doing well despite 11 medicines between them.

Brittney graduated with an Associates degree in Business at the College of DuPage in Glen Ellyn, IL and a B.A. in Business Management from North Central College in Naperville, IL. She has eight years cumulative work experience in Banking & Finance. Since having her first child five years ago, she has stayed home taking care of her beautiful, yet "medically challenged" children. She has volunteered in a number of settings. Currently, she is working on her church's counting team to process the offerings.

Elva Smith

Elva was a CRN Committee Member working on the Research Committee. She will continue her work on the Research Committee as a Board member. Elva is a 71 year old grandma and a native of Georgia. She was born in Savannah and raised in Atlanta. She is an ASCP registered medical technologist, and has worked, off and on, for over 50 years in various clinical laboratory settings. Her last employment was with the Centers for Disease Control and Prevention in Atlanta.

She is retired and a widow with 4 children, and 5 grandchildren. Her grandson, Mitchell Smith, was diagnosed with cystinosis in November 2004 at 17 months and is now just over 3 years old. Elva is thankful that Mitchell has responded well to his medications and is a typical little boy full of energy and mischief. He lives in Lawrenceville, GA with his 4 year old sister Rachel and parents, Eric and Lorna Smith.
Cystinosis Research Update
By Christy Greeley, Vice President, Research

2006 Research Funding Announced

The CRN Scientific Review Board met in July 2006 to review the studies received from this year’s call for proposals. We are proud to announce that two grants have been funded as a result of the SRB’s recommendations. They are:

“Study of ATP metabolism in human cystinotic proximal tubular cells and in humans with cystinosis in vivo”
Elena Levchenko, M.D., Ph.D., University Medical Center Nijmegen, The Netherlands
Grant Timeline: January 2007 – December 2007
Award Amount: $68,090

Cystinosis is an autosomal recessive disorder with multi-organ involvement. The disease is caused by mutations in the CTNS gene, which maps to 17p13 and encodes the lysosomal cystine carrier, cystinosin. Cystinosis initially affects the kidneys and subsequently other organs including eyes, central and peripheral nervous system, muscles, endocrine organs and liver. The aminothiol, cysteamine, depleting intralysosomal cystine, retards the deterioration of renal function and postpones the occurrence of extra-renal complications. This is in agreement with increased lysosomal cystine levels being the primary cause of this disease. However, the subsequent steps in the pathogenesis of cystinosis are unknown. Alterations of intracellular energy metabolism were postulated to be an underlying mechanism of cellular dysfunction in cystinosis based on the finding of ATP depletion in proximal tubular cells loaded with cystine dimethyl ester. However, this mechanism was neither confirmed in human renal material nor in humans with cystinosis. The study of ATP metabolism in human cystinotic proximal tubular cells and in humans with cystinosis and controls is the subject of this research proposal.

The project is based on (i) our initial observations that cystinotic fibroblasts and granulocytes display a significant decrease in intracellular ATP and (ii) development of differentiated human cystinotic proximal tubular cell lines, carrying known CTNS mutations, which cystine accumulation approaches the in vivo situation.

Apart from this we will evaluate intracellular ATP status in vivo in patients with cystinosis using MR spectrometry.

Therefore, the current study consists of both a fundamental part and a clinical part.

A. The fundamental part focuses on:

The investigation of ATP homeostasis in temperature sensitive immortalized proximal tubular cell lines from cystinotic patients and healthy controls at different stages of cystine accumulation and after exposure to cysteamine.

B. The clinical part focuses on:

Measurement of ATP status in vivo in patients with cystinosis.

The demonstration of altered ATP homeostasis in human cystinotic proximal tubular cells and in humans with cystinosis will set a major step towards better understanding the pathogenesis of the disease and ultimately to develop new therapeutic strategies in patients with cystinosis.

continued on page 8
“Development of a Cysteamine in situ gelling system for the topical treatment of corneal crystals in Cystinosis”

Dr C Tuleu, University of London, School Of Pharmacy, Dr Olufemi Rabiu, Guy’s & St Thomas’ NHS Foundation Hospital, Mr Ken K. Nischal, Mr Rajnish Sekhri, and Dr William Van’t Hoff, Great Ormond Street Hospital and Institute of Child Health

Grant Timeline:  Fall 2006 – Fall 2009
Award Amount:  $192,660

To produce a once daily delivery system for topical ocular cysteamine in patients with cystinosis. The overall aim of this project is to develop and test a new formulation of topical cysteamine. The rationale is that increased ocular contact time of the drug using an in situ gelling and mucoadhesive polymer will reduce lacrimal drainage and thereby increase bioavailability. This should lead to decreased frequency of administration which should greatly improve compliance. The first aim will be to formulate a cysteamine ophthalmic preparation in a polymeric vehicle. This will be tested in vitro and then in vivo evaluation will be undertaken in rabbits prior to clinical testing in patients with cystinosis.

CRN is very excited to support these international investigators, both of whom are receiving funding from CRN for the first time. We will keep you updated on their studies’ progress.

In 2006, CRN supported six studies which account for nearly $600,000 in research funding.

CRN Funds Dr. Trauner’s Team to Attend Netherlands Conference

CRN funded a special request from Dr. Doris Trauner, neurologist at UCSD, for four of her testers to attend the International Cystinosis Conference in Amsterdam this past summer. The study is entitled "Cognitive Profiles in an International Sample of Individuals with Cystinosis" and represents a one time grant of $12,500. Excerpts from the grant are included below:

1. Abstract/Summary of project

We are requesting funds for travel and accommodations for testers from Dr. Trauner’s Pediatric Neurology Research Group to attend the International Cystinosis Conference in Amsterdam (June-July 2006) in order to collect cognitive data on international participants. The goal of this project is to test at least 20 international participants so that we may compare the cognitive profiles of our North American sample of cystinosis individuals with an international sample. Results will be combined with data collected in Italy in 2000 and will be written up for publication in a scientific journal.

2. Hypothesis and specific aims

Cognitive testing on individuals with cystinosis in North America has shown a specific pattern of performance with better verbal than non-verbal skills. We plan to examine cognitive skills in an international group of individuals with cystinosis to determine whether this pattern is seen in individuals of other origins or is specific to our North American sample.

We currently have data on 5 international participants who were seen at the 2000 International Cystinosis Conference in Italy. It would be extremely valuable to collect additional data on an international sample so that formal analyses of the data can be conducted.
Cystinosis Research Update

Based on our preliminary data from the Italy conference, it is hypothesized that the specific cognitive pattern seen in North American subjects is also present in the international sample. This suggests that the pattern is directly related to cystinosis rather than being a product of the “culture/environment” (e.g., differences in the educational system, treatment approaches, or cultural values).

3. Methodology

We plan to conduct psychometric testing on individuals with cystinosis ages 4 years to adults. We have a standard test battery that has previously been used with an international sample, consisting of numerous non-verbal tasks. For individuals who do not speak or understand English, test instructions and any verbal tasks will be translated by a parent or other conference attendee on a voluntary basis. The testing will take approximately one 3-hour session that will be scheduled at each participant’s convenience. The testing is non-invasive and behaviorally-oriented, and it helps us learn more about the cognitive and behavioral effects of cystinosis. Individuals will be asked to perform such tasks as putting together puzzles, defining words, and identifying or remembering pictures. Participation in the psychometric testing will be available to individuals with cystinosis (children and adults) attending the conference and will be entirely voluntary.

Research Update from Dr. Antignac

The following one year progress report was submitted by Dr. Corinne Antignac:

CRN PROGRESS REPORT – SEPTEMBER 2006
Development, and in vivo testing, of novel therapies for cystinosis

Our 2 yr research project is a joint proposal between:

Dr. Corinne Antignac (Inserm U574, Hôpital Necker-Enfants Malades, Paris, France) - Project 1

Dr. Vasiliki Kalatzis (CNRS UMR 5535, Institut de Génétique Moléculaire, Montpellier, France) - Project 2.

The money allotted to Project 2 is currently being used to finance Claire Hippert, a Ph.D. student.

Progress report Project 1 – Cysteamine trial

i) Characterization of the renal phenotype in Ctns-/mice backcrossed onto pure genetic backgrounds

Prior to treat Ctns-/mice with cysteamine to verify its efficiency, we have performed extensive analysis of the murine model of cystinosis. The identification of the CTNS gene enabled us to generate a mouse model of cystinosis by disrupting the murine homologous of CTNS (Cherqui et al., 2002). This model has been shown to present some of the findings in patients with cystinosis, such as cystine accumulation, cystine crystal deposition in tissues and ocular anomalies. However, these mice do not develop any renal abnormalities, aside from rare vacuolised renal tubules in a small number of mice.

Knowing that the genetic background can strongly influence the phenotypic severity, we transferred the Ctns mutation from our mixed C57BL6/129sv background project onto pure C57BL/6 and FVB/N backgrounds. We then verified that the mice were accumulating cystine in their organs. In addition, we performed various histological studies, which interestingly showed that whereas FVB/N Ctns-/- mice do not have renal lesions, C57BL/6 Ctns-/- mice present several histological findings. Chronologically, the first lesions we observed were vacuolised tubules, which appeared around 6 months of age. At 9 months, proximal tubular atrophy or cellular infiltration was found. Notably, these lesions were focal; hence, both normal and affected areas were seen in the same kidney. We initially measured serum and urine parameters and failed to see any global differences in renal function between Ctns +/+ and Ctns-/- mice. Given that the lesions do not involve the entire kidney in Ctns-/- mice, these measures of global renal function may fail to detect a mild disorder in renal function. Thus, we plan to study tubular function more precisely in these mice.

This murine model of cystinosis, which develops renal lesions, will be a relevant model on which the cysteamine trial can be performed.

continued on page 10
(ii) Immortalised cell lines
With the goal of establishing relevant cell models to study cystinosis, we are currently generating several tubular cell lines.

Last year, we mated Ctns mice with the Immortomouse, bearing a thermosensible mutant of the SV40 T antigen in their genome (Jat et al., 1991), and isolated proximal tubular cells from 6 Ctns-/- and 5 Ctns+/- mice. The cells we isolated were conditionally immortalized and could either proliferate or differentiate when grown at 33° or 39°, respectively. We then demonstrated that the Ctns-/- cell lines accumulate cystine and that all the cell lines exhibit γ-glutamyl transpeptidase activity consistent with a proximal tubular phenotype. This year, we further characterised these cell lines. First, we detected alkaline phosphatase activity in these cells. Furthermore, when the cells were grown on filters, transepithelial resistance (around 50 Ω/cm²) was measurable, demonstrating that the cells are polarised. Additionally, we observed that α-tubulin, β-catenin and zona occludens 1 were properly localised in these cells; β-catenin and zona occludens 1 were detected at the plasma membrane, whereas α-tubulin was found in the submembranous region. The results of all these studies are consistent with the cells possessing a proximal tubular phenotype. At present, we are performing biochemical analysis to evaluate the redox status of these cells.

The human cell lines we previously generated were permanently immortalised and these cells mildly express tubular markers. Thus, we aimed to generate human tubular cells that are conditionally immortalised. We obtained cells from the urine of 3 cystinosis patients and our collaborators at the Centre de Thérapie Génique (Nantes, France) transduced these with a thermosensitive SV40 T antigen. We have recently received these new cell lines and we will now perform the same sets of experiments to verify the proximal tubular phenotype of these cells. In addition, we also generated 2 control tubular lines from normal individuals.

(iii) Antibodies against cystinosin
Since polyclonal antibodies generated against cystinosin-derived peptides were highly batch-dependant and since no reliable antibodies directed against the N-terminal part of cystinosin could be generated by this approach, we decided to use the Baculovirus expression system to produce recombinant cystinosin proteins in insect cells. This system has the unique advantage of producing a high level of recombinant proteins that can undergo several post-translational modifications, which is crucial for cystinosin as the N-terminal part of the protein is highly glycosylated.

We have already generated a CTNS transfer vector by inserting the 5’ part of the CTNS gene (encoding amino acids 24 to 123) preceded by sequences encoding a signal peptide and a 6XHIS tag in the multiple cloning site of pFastBac1. This vector is currently used to produce the recombinant protein with the Bac-to-Bac® Baculovirus Expression System (Invitrogen), which will then be injected to rabbits to produce polyclonal antibodies [in collaboration with Dr Y. Boublik, at the CRBM (Montpellier, France)].

Progress report Project 2 – Gene transfer studies
We have made significant progress in two parts of our original research proposal.

i) Validation of in vitro gene transfer studies
To validate our in vitro gene transfer studies showing that the efficiency of cystine reduction is age-dependent in primary murine hepatocyte cultures, we began tail vein injections of human adenovirus vectors expressing CTNS (hAd-CTNS) or CTNS fused to the green fluorescent protein GFP (hAd-CTNS-GFP) to target the liver in different aged Ctns-/- mice. Initially, we performed liver biopsies on Ctns-/- mice on day 1 (n = 9), injected the mice with hAd-CTNS (n = 3), hAd-CTNS-GFP (n = 3) or hAd-GFP (n = 3) on day 2, and sacrificed the mice on day 9. However, the liver biopsy itself caused a reduction in cystine levels likely, due to a regeneration of the liver. We thereby altered our experimental plan to compare cystine levels post-transduction with those of non-injected mice: we injected mice (n=9) with hAd-CTNS (n = 4), hAd-CTNS-GFP (n = 4) or hAd-GFP (n = 4) on day 1, and sacrificed the animals on day 8; we used 4 non-injected age-matched Ctns-/- mice and 3 Ctns+/- as controls.
Following sacrifice, we evaluated the liver transduction efficiency by FACS analysis (GFP expression) and by immunofluorescence studies (GFP expression or using an anti-cystinosin antibody). We evaluated cystine clearance using the radio-competition cystine binding protein assay. Following injection of 9 month-old mice, we did not reduce cystine levels with any vector 1 week post-injection despite a 70% transduction efficiency. In contrast, following injection of 2 month-old mice, we reduced cystine levels with the hAd-CTNS vector despite a lower transduction efficiency. We are currently repeating the injections of 2 month-old mice to confirm these data and we will also inject an intermediate age (6 months).

### ii) Eye gene transfer

Our *in vitro* data suggests that cystine reduction is age-dependent, thus a spatial and temporal guide of the ocular anomalies in the *Ctns*<sup>−/−</sup> mice is a necessary prelude to *in vivo* gene transfer studies. We recently finished the biochemical, histological and clinical characterisation of the ocular anomalies in *Ctns*<sup>−/−</sup> mice.

**Biochemical assay:** We dissected the mouse eye into cornea, iris (plus ciliary body), neural retina and lens, and analysed the cystine levels in each tissue versus age (1, 3, 5, 7, 9, 11, 13, 15-17, 19-21 & 23-25 months). We detected elevated cystine levels in the iris (including the ciliary body) of *Ctns*<sup>−/−</sup> mice from 1 month compared to wild-type mice, and in the cornea and retina from 3 months. For the lens, a significant difference was seen from 5 months. The comparison of cystine accumulation for different *Ctns*<sup>−/−</sup> tissues showed that cystine levels in the cornea and iris were globally the highest and increased at a greater rate with age. Cystine levels increased dramatically between 5 and 7 months, peaked at 9 months with levels 90- and 260-fold over wild-type for the cornea and iris, respectively; cystine levels were relatively stable from thereon. By comparison, retinal cystine levels increased less dramatically with age and at 9 months were 70-fold over wild-type. Finally, the lens contained the lowest cystine levels: 6-fold over wild-type at 9 months and reaching a maximum of 30-fold at 23 months.

**Histological study:** We screened for the presence of cystine crystals and lesions versus age (1, 3, 5, 7, 9, 11, 13, 15, 19 and 22 months). We detected rare cystine crystals in the iris and ciliary body from 1 month. The number of crystals increased with age and became abundant from 7 months. Moreover, in 19 and 22 month-old *Ctns*<sup>−/−</sup> mice, we noted a fusion of the iris and cornea. In contrast to the iris and ciliary body, we did not detect corneal crystals at 1 month of age. The corneal crystals appeared from 3 months and were located within the keratocytes and distributed throughout the corneal stroma. We did not detect cystine crystals in the epithelium, Bowman’s layer, endothelium or Descemet’s membrane. Finally, in addition to the anterior synechiae, a vascularization of the cornea was also visible from 19 months.

In the choroid and sclera, we first detected cystine crystals from 3 and 5 months, respectively, and their number also increased with age, but at a slower rate. In addition, from 7 months the choroid of *Ctns*<sup>−/−</sup> mice was thinner and less homogenous than that of *Ctns*<sup>+/+</sup> controls and continued to degenerate with age. Up to at least 15 months, we could not detect crystals in the retina and both the neural retina and the retinal pigment epithelium (RP) retained a homogeneous appearance in *Ctns*<sup>+/+</sup> and *Ctns*<sup>−/−</sup> mice. In contrast, in the 19 and 22 month-old *Ctns*<sup>−/−</sup> mice, the RP showed signs of degeneration. Moreover, the photoreceptor segments and nuclei were absent and the inner nuclear layer constituted the most posterior layer of the neural retina and contained rare crystals. Finally, we could not detect cystine crystals in the lens at any age.
Cystinosis Research Update

continued from page 11

Taken together, the appearance of cystine crystals is consistent with the data we obtained from the cystine assay: i) we did not detect cystine crystals in the lens, consistent with the low cystine levels in this tissue (maximum of 2 nmol 1/2 cystine/mg protein); ii) we found rare retinal crystals in the oldest mice when cystine levels reached ~30 nmol 1/2 cystine/mg protein, and iii) we detected cystine crystals in the iris and ciliary body prior to 5 months, however, these became abundant from 7 months, consistent with the sharp 3-fold increase from 30 to 80 nmol 1/2 cystine/mg protein seen at this age. Furthermore, the biochemical data provides an insight into the cystine levels of different eye tissues, which to our knowledge is not known in cystinosis patients, whereas the histological data are consistent with those reported in patients.

Clinical tests: We used four different tests to determine the biological effect of cystine accumulation.

i) Slit lamp photography: We examined the evolution of cystine crystals in the cornea and iris with age (1, 3, 5, 7, 9 & 13 months). We first detected rare crystals at 3 months in both tissues and, consistent with the histological data, crystals were easily detectable and abundant from 7 months. Thus, although corneal crystals appeared slightly later, their evolution paralleled that of the iris, consistent with the overlapping profile of cystine levels in these tissues.

ii) Intra-ocular pressure (IOP): Differences in IOP can be due to obstructed flow of aqueous humour (increased IOP) or defective production by the ciliary body (decreased IOP). Therefore, we indirectly examined the effect of ciliary body crystals on humour production by measuring IOP. We measured the IOP of 6 Ctns+/+ and Ctns−/− mice aged 15-19 months using a Tonopen XL and found that the IOP of the Ctns−/− mice was significantly decreased. These results suggest that the presence of ciliary body crystals could disrupt aqueous humour production and account for this relative decrease in IOP.

iii) Electroretinogram (ERG) profiles: We studied the retinal function of Ctns+/+ and Ctns−/− mice as a function of age (3, 5, 7, 9 & 11 months). We did not detect a significant difference in a-wave or b-wave amplitude between the Ctns+/+ and Ctns−/− response at any age. Taken together, these data suggest that retinal function is preserved in Ctns−/− mice up to 11 months, consistent with the low cystine levels and the absence of retinal cystine crystals.

iv) Light/dark exploration test: We assayed 17-19 month-old Ctns−/− mice for signs of photophobia. Both the Ctns+/+ and Ctns−/− mice exhibited a clear preference for the dark compartment, where they spent almost two thirds of the duration of the session. The Ctns−/− mice, however, showed an increase in the total time spent in the light compartment, but not in the number of entries into the light section or the mean duration of each visit. Interestingly, the increased total time the Ctns−/− mice spent in the light section was due to an increased number of short-duration (inferior to 10 s) visits, which, on average, lasted 5.1 ± 0.3 s. Thus, although a classic photophobic behaviour was not detected, our results highlight a need for the Ctns−/− mice to rapidly re-enter the dark compartment significantly more often than the wild-type mice, which could suggest a mild photophobic state.

Taken together, this temporospatial guide of the ocular anomalies of the Ctns−/− mice, in addition to validating the mouse model, provides a foundation for the future testing of novel therapeutics to efficiently restore a normal ocular phenotype. We are now in a position to continue with the next part of our research proposal, eye targeted in vivo gene transfer studies.

Please note that this work is documented in our manuscript: “A temporospatial guide to the ocular anomalies in a cystinosis mouse model” by V. Kalatzis, N. Serratrice, C. Hippert, O. Payet, C. Arndt, C. Cazavieille, T. Maurice, C. Hamel, F. Malecaze, C. Antignac, A. Müller and E.J. Kremer, which is submitted for publication.

continued on page 13
Dr. Thoene’s Tissue Repository Study Begins

CRN is pleased to announce that funding has begun for Dr. Jess Thoene’s study, "Tissue Repository for Cystinosis". As you will recall, this study was approved at the 2005 Scientific Review Board meeting, but was put on hold when Dr. Thoene was displaced from his Tulane laboratory as a result of Hurricane Katrina. Dr. Thoene is now back at the University of Michigan in Ann Arbor.

Below is a summary of the protocol:

1. Abstract and Summary:
Research on rare diseases is hampered by unavailability of tissue. This problem was noted by the National Commission on Orphan Diseases and has been addressed in various venues. A tissue repository for samples from patients with nephropathic cystinosis has not yet been established. Our current inquiries into the nature of cell death in cystinosis, specifically enhanced apoptosis (1,2) which we have demonstrated in tissue culture, along with the demonstration of a tubular glomeruli in a kidney removed from a cystinosis patient, makes the situation now appropriate for action. I propose to obtain blocks of renal tissue obtained at biopsy for clinical indications from patients with cystinosis (nephropathic and intermediate) and store them coded, along with a brief clinical summary.

CRN Financial Update

Cystinosis Financial Update
By Brittney LeBeau, Treasurer

I would like to take this opportunity to convey how excited I am to assume the position of Treasurer for the Cystinosis Research Network. My husband, Brett, and I have two children with cystinosis, Alexis and Evan. For many years, we have benefited directly from the efforts of a group of individuals in our community focused on providing us assistance in our moments of need. We have always contributed in support roles and in our own way. When I was asked if I would consider taking the position of Treasurer for the CRN, I was truly touched. I realized it was my time to step forward as a leader and expand our level of giving back. More specifically, my decision came as I felt a need to help give back to those who have helped me so much in the CRN.

I will be assuming the Treasurer responsibilities from Colleen Hammond. For those of you who know Colleen, you are very aware of all of the contributions and sacrifices she has made in not only creating our organization but also providing the necessary leadership. After more than 10+ years of sacrifice and dedication, Colleen Hammond has officially stepped down as the CRN Treasurer. We are very grateful for her contributions to the CRN and wish her the very best as she focuses on her family’s immediate needs. Of course, we all know Colleen is always just a telephone call away!

With some big shoes to fill, I have decided to take on this new and exciting challenge. I have more than 8 years of banking and finance experience under my belt, and I feel very qualified for the position of Treasurer. Planning for the transition of responsibilities has started, and it will take several months to complete. There is no specific date set, but this is a great opportunity to streamline many parts of the Finance Committee’s duties and the programs being used, both externally and internally. So stay tuned!
Spotlight on Dr. Ewa Elenberg

Dr. Ewa Elenberg sits on the Medical Advisory Committee for the Cystinosis Research Network. She recently spoke at a medical conference in Poland and shared thoughts that cystinosis families had written to her. Her presentation was well received. We asked her to tell us how she became interested in cystinosis and subsequently involved in the CRN. Here is her response...

By Dr. Ewa Elenberg

My interest in cystinosis was sparked by meeting Shea Hammond and his family.

In the summer of 1994, during one of my first night calls as a Nephrology Fellow in Boston, I was called by Shea's mom telling me that her 4 year old son has Cystinosis and now is vomiting too much, and he needs to be admitted to the hospital and started on iv fluids. I was surprised to be given a plan by the mother, without even waiting for me to make any decisions. On the other hand, while I vaguely knew the name of this disease, I had never seen a patient with Cystinosis, and I did not really have any better ideas. Colleen waited for a few seconds, giving me time to think and then asked me a straightforward question: "Do you know anything about Cystinosis"? Her question was so direct and quick, that with the sense of guilt I gave a very honest answer: "Actually, I don't..." I admitted to having read only a few sentences on the disease in a pediatric textbook. I put her on hold, ran to pick up a book from the shelf, found the chapter, but other than already recalled few sentences, I did not find much information. Colleen was very kind and patient with me. She said that she was very happy that I was honest and briefly outlined for me the major problems with this disease and how to deal with them. She told me that she would be happy to share with me some more information the next morning. Indeed, the first articles I read on Cystinosis were those received the next day from Colleen.

When I entered Shea's room the next morning, the room was dark and extremely cold, the boxes of salt pretzels were everywhere. What I saw puzzled me, so I asked why it was so cold and Colleen answered that Shea liked it that way, because this helped with his nausea. Darkness eased his photophobia. The pretzels - did not make sense to me, as I was told that Shea was not eating anything and was totally dependent on total parenteral nutrition.

Dr. Ewa Elenberg

Current Position
Assistant Professor of Pediatrics
Department of Pediatrics, Renal Section
Baylor College of Medicine, Texas Children's Hospital

Medical Education
M.D. Medical University, Wroclaw, Poland

Postdoctoral Education
Educational Scholars Fellowship Program,
Baylor College of Medicine, TX

Professional Memberships
American Academy of Pediatrics
Cystinosis Foundation
National Kidney Foundation
Cystinosis Research Network
American Society of Nephrology
International Pediatric Nephrology

Languages
Fluent in: English and Polish
Conversant in: Spanish and German
Knowledge of: French, Latin, and Greek

Recent Lecture
June, 2006 37th Annual Conference of the European Working Group on Psychosocial Aspects of Children with Chronic Renal Failure. Presentation was titled, “Cystinosis-the Challenge of Living with a Chronic Illness.”

Upcoming Lecture
January, 2007 Cystinosis Mexico’s 4th Annual Cystinosis Conference

Continued on page 15
Spotlight on Dr. Ewa Elenberg

Continued from page 14

So I expressed my surprise and said, "Wow, you must be really crazy about these pretzels" and Colleen answered, "Oh, no, these are for Shea". This really did not make sense to me, so Colleen took some pretzels, gave one by one to Shea who licked all salt and discarded the rest, just to take another one to his mouth and to lick same way as the previous one. Then Colleen said, "This is what I call eating..." His inability to eat was so puzzling to me that I ran to the library to do a literature search but I found no information on eating problems in Cystinosis. This episode spurred my interest in finding out if there are more patients like Shea who have eating problems. I designed the survey and started calling some of the parents of children with Cystinosis, parents who knew Colleen and who were known to be supportive of research. I started collecting data and soon realized that eating problems were very common in Cystinosis, though not yet described in the literature. The more I interacted with the families over the phone, the more I became interested in this disease and the more I admired the families who really did not have much medical and psychologic support. Everyone was eager to talk to me and did not complain that I was calling in the evening to answer the survey. I called one mother from Canada close to 10pm and when I found out that she was admitted to the hospital while being in labor, I tried to end the interview, but she wanted to answer my questions. She was very nice and enthusiastic and she was happy that I was asking her about the eating issues that she always felt her child had.

This research project led to the initial description and to better understanding the spectrum of feeding problems in Cystinosis. It allowed me also to design the plan to help Shea learn to eat. I worked together with Shea’s mother trying to find methods of introducing a food to Shea as a part of daily routine. Initially we decided to use food as a play tool, allowing him to play with spoon, plates, food. Every evening he was sitting with the family at the table playing with his tomato pasta plate or soups spreading all around the table while the whole family pretended that nothing was happening. Shea was learning that food was a part of family life. I found out that Shea likes ice cubes, so I asked Colleen to freeze chicken broth and serve him as ice cubes. He liked it! Then we started experimenting more and more... Slowly, month after month we were able to see a small improvement, after 2 years he was completely weaned off intravenous nutrition. Now Shea probably does not remember his past problems. He is 17 years old, a recent renal transplant recipient who enjoys eating!

Since meeting Shea, I got to know many of you at various conferences. I have no words to express the depth of my admiration for you. You not only deal with daily issues of living with a complex chronic illness, but you also have energy to spend countless hours preparing the Cystinosis Symposia, taking your precious time off work to meet, to educate each other, to build up the strength fighting many obstacles. Every day is a new challenge, every day is a strenuous routine ruled by the clock. Your life is divided into hours, always remembering which medication to take or to give. Time without vomiting or discomfort is considered a "happy time". Since I met you, you have always been smiling, never complaining, never angry with the lack of knowledge by the medical staff. On the contrary, you are eager to teach all who want to learn, you are supportive of each other and doctors as well. You don’t assess blame but try to help us to understand the scope of issues and how to deal with them. You join the force together trying to "beat THIS THING", with dignity and positive thinking, with hope, not anger and despair... I cannot find better words to describe how I admire you. You, the patients and the families are my HEROES.
Update from Cystinosis Mexico AC

By Victor Gomez
Cystinosis Mexico AC

We would like to thank the Cystinosis Research Network for letting us inform the cystinosis community of how our association is working in Mexico City.

Cystinosis Foundation Mexico was born in January, 2002. Our goal is to help and support patients with cystinosis in Mexico. The first stage was done. In 2006 we are starting the second phase, Mexican Association of Cystinosis. We are establishing ourselves as main resource for cystinosis in all of Latin America.

We are pleased to announce that our 3rd family-medical conference will be held in Pachuca Hidalgo (town located 1 hour from Mexico City). Our major sponsor will be “D I F “ (Mexican Institution dedicated to help financially poor families, scholarships and homeless kids). We will inform conference attendees of our new projects and strategies. Some of them are:

- Support to South American patients with cystinosis working together with CRN
- 2nd Program to search for patients with cystinosis around Mexico and to create general cystinosis awareness in Mexico
- Acquiring complete medical assistance for Mexican patients with cystinosis

National Institutes of Pediatrics in Mexico also is hosting the 1st Cystinosis research mutation program in the Mexican population.

These are some of the main projects which will start in January, 2007. Some of these programs will be sponsored at first by American cystinosis groups & laboratories. Stay tuned. You will soon see more details on www.cystinosismexico.org in English & Spanish.

Cystinosis Mexico would like to say THANK YOU and CONGRATULATIONS to the Cystinosis Research Network on its 10th anniversary! CRN has done great work for ALL patients with cystinosis and their families! Results can be seen by the many research programs they are currently supporting, the Internet support group, and the successful conferences they have organized.

Dr. Belmont, Medical Director, Mrs. Victoria Ruffo, President DIF Municipal Pachuca, and Víctor Gómez, President Cistinosis México
Kelly Bryant Receives 2006 CRN Scholarship

CRN is pleased to announce the recipient of the 2006 Cystinosis Research Network Scholarship, Ms. Kelly Bryant from La Crosse, WI. Ms. Bryant maintained a 3.8 GPA (on a 4.0 scale) in a rigorous college preparatory curriculum at Central High School in La Crosse.

In addition to her academic distinction, Kelly has been an active volunteer, contributing many hours to Gundersen Lutheran Medical Center, the Salvation Army, and as a food server for the Place of Grace. She has been accepted at the University of Wisconsin—River Falls, where she will be attending in the Fall of 2006, majoring in journalism. Her professional goals are to become a journalist and work either in the print media as a reporter or to write for a website. Congratulations, Kelly!

Jack Kitchens Featured on ESPN BassCenter

Jack Kitchens caught his first bass, and it is well documented! After he caught the bass, his dad, Travis e-mailed a photo of Jack and the Bass to ESPN2's BassCenter. The show has a segment called "Destination: Bass." Each week they post someone's photo with his/her catch. Travis e-mailed the photo on late Friday afternoon. BassCenter liked the photo so much, they bumped what they had scheduled to put on Saturday and used Jack's picture instead!

John Kernan, one of the show's host, told Jack's story like this, "Jack picked the bait, picked the spot, and was working the trolling motor when he caught the bass! Congratulations to Jack for his first solo bass."

Travis said he mentioned cystinosis in the e-mail to BassCenter, but they were unable to mention it on the air. They said they may very well contact the Kitchens' in the future to do a story on Jack and cystinosis.

Jack is six years old and was diagnosed with cystinosis at age 13 months. He lives in Georgia with his Mom (Bette), Dad (Travis), and little sister, Ava.
Megan Morrill Wins at West Coast Horse Show

By Jill Morrill

Megan Morrill, a 10 year old with cystinosis from Grand Rapids, MI, is passionate about horses. She recently won the West Coast Horse Show Equestrian Events for Jumper and Equitation. Megan has tried many sports, such as soccer, dance, T-ball, gymnastics, and swimming but has chosen horse back as her #1 sport.

Megan began riding 3 1/2 years ago at Kentree Stables and has never looked back. She is an animal lover so horses seem a very natural fit. Megan enjoys the barn so much that even when she is not feeling 100%, she still wants to go to the barn. The horses only seem to cheer her up. Megan has been showing horses for two summers, and this past summer, she accumulated the most points in her division to win the summer championship event. She jumps fences and is fearless. She is firm with the pony she shows but also very gentle and tender. She has found amazing confidence and true joy in her life because of horseback riding.

Her parents, Jill and Brian Morrill, couldn't be happier or more proud of her. She also has a younger brother, who is also proud of her (but a little jealous of her big trophy and ribbons!) Most of all, the Morrills are thrilled with the quality of life Megan has because of her wonderful doctors, researchers, and the Cystinosis Research Network.
A Tale of Two Transplants

Andrew Hoffman and Shea Hammond are two typical teenage boys. No one would know that they both have cystinosis and have both had kidney transplants. Andrew and Shea received their transplants less than a year apart from one another. They both received them at the National Institutes of Health (NIH), where they were both part of the steroid-free protocol. Andrew received a cadaver kidney, and Shea received a kidney from a living related donor. Both stories have similarities and differences. Their families took a moment to share their stories...

Andrew Hoffman

By Don, Doretta, Jon, and Andrew Hoffman

Andrew was diagnosed with cystinosis in early 1989 when he was 10 months old. Since diagnosis, Andrew’s life has been followed closely by mainly Dr. Gahl at the National Institute of Health (NIH), Dr. Woodhead, Dr. Porter, and now Dr. Lee at the University of Iowa for Andrew’s pseudotumor cerebri—four very wonderful and dedicated doctors!

During Andrew’s August, 2004, regularly scheduled checkup at the NIH, we were told that Andrew’s kidneys were starting to deteriorate and we should begin to prepare ourselves for a transplant in the near future.

Dr. Gahl informed us of the protocol study for steroid free transplantation that was being preformed at the NIH. Andrew’s thinking was he had been under the care of Dr. Gahl and his team for 16 years, and the NIH is where he wanted to have his transplant - when the time came.

In late September, 2005 we decided to put Andrew on the donor list through the NIH in Maryland. During this time, Doretta was finishing testing to determine if her kidney would be a match for Andrew. The transplant from the donor list or Mom was scheduled for anytime in December.

Shea Hammond

By Colleen Hammond

In many ways, Shea Hammond is a typical 17-year-old high school junior. He greatly enjoys the freedom of having his license and his own car. He goes to the gym after school to lift weights. He spends most weeknights doing hours of homework, as he works to get good grades that will help him to get into the college of his choice. He spends weekends with his friends going to movies and playing basketball. Yet, in many ways, Shea has led a very different life than most of his peers. He spent the past summer at the National Institute of Health getting a kidney transplant and he has spent his life courageously battling cystinosis and refusing to let it control his life.

August 15, 2006 marks a sort of rebirth for Shea. When Shea received a new healthy kidney, his quality of life greatly improved. Shortly before the transplant, Shea was feeling the awful affects of kidney failure. His creatinine had quickly risen to 10 and he was feeling tired, weak and “foggy.” The wonderful transplant team at NIH led by Dr. Mannon and Dr. Kirk worked quickly so Shea could have the kidney transplant without going on dialysis. Shea selected the NIH transplant protocol for people with cystinosis because he felt that it would provide him with the best quality of life.

A good quality of life has always been the highest priority for Shea. He has always faced each day with a
Andrew, continued from page 19

At 9:45 a.m. on Oct. 3, Don’s birthday, we received a call from Liz Hale, transplant coordinator at the NIH. Hale relayed over the phone there was a kidney available for Andrew, and we should come to the NIH as soon as possible.

We made plane reservations through our tremendous travel agent (Barb), called family members, packed our bags (dirty clothes and all), and we were off to the NIH!

We arrived at the NIH at 7:47pm on Oct. 3, and Andrew, now 17 years old, had his transplant at 6:00 a.m. Oct. 4th under the care of Dr. Kirk and Dr. Hale. The transplant was a textbook surgery with no complications. After the transplant, we were informed that the donor kidney was a perfect match for Andrew and was a better kidney than Doretta could have given Andrew. We believe that this is truly a gift from God. Andrew remained in the hospital for seven days and stayed at the Children’s Inn for two more weeks, and then we were on our way home.

On October, 24th – 21 days later – Andrew arrived back in Iowa to continue a steady recovery accompanied by a slowly decreasing number of medications. We received tremendous support from friends, family, and the community.

Since the transplant, everything has been going very well for Andrew.

Our advice to our other cystinosis families is that the NIH is a wonderful place. The doctors, transplant coordinators, and nursing staff are wonderful people and work so hard to make sure that everything is taken care of and running smoothly. Our family can not say enough about our positive experience at the NIH. These are some of the best doctors and health professionals in the world and do so much to help cystinosis families. We can not begin to express how much they mean to us. We now call them family, and what a wonderful family to have!

Thank you to Heidi Hughes and Morgan Friend for paving the way for cystinosis patients like Andrew to receive steroid free transplants at the NIH. If anyone has any questions please feel free to contact us at (515) 733-4437. We will glad to answer them.

Shea, continued from page 19

wonderful attitude of humor, joy and gratitude. Shea faced the challenges of the kidney transplant with the same positive attitude. As a family, we followed the lead of the NIH transplant team of nephrologists, surgeons, nurses, and dieticians. Each member of the family had a role to play. Shea and his dad, Jack, set the teams. The boys’ team was called the “Shacks” – Shea as open receiver and Jack as his caregiver. The girls teams was called the “Kalleens” – Kayla (Shea’s 19 year old sister) as caregiver and me as the organ donor. Kayla and Jack were tremendous caregivers in every way. When Kayla learned Shea would need a kidney transplant over the summer, she insisted she cancel all of her summer plans, and go to the NIH with us. Kayla has always been Shea’s best friend and has willingly sacrificed much to help Shea. When Kayla and Shea talk about their summer, they say that although it was difficult for Shea to have the transplant, they greatly enjoyed spending the summer together before Kayla left for her freshman year at Harvard.

We are so thankful to the entire NIH transplant team and staff at the Children’s Inn. The transplant process has been a great experience. The care provided by the team is excellent on every level. They meticulously addressed all of Shea’s medical needs and also his psychological needs related to being a young man going through the transplant.

I was the donor and had laparoscopic surgery. My surgeon, Dr. Pinto, was fantastic. I compare the transplant process to that of having a baby. It is one of the most miraculous and joyful events in your life that you will ever experience. It can be a roller coaster ride of emotional ups and downs. However, in the end, you forget the pain and focus on the end result - a healthy child.

We are very grateful to our family and our cystinosis family for all of their support. Thank you Heidi Hughes, Morgan Friend, and Andrew Hoffman and families for paving the way for us by having transplants at NIH before Shea. Andrew was at the NIH when Shea had his transplant. He and his father, Don, were incredibly helpful in answering all of our questions and helping to dissolve any fears we had.

Shea feels better than he ever has in his life. He has tremendous energy and greatly improved ability to focus on schoolwork. He is grateful to his health care team of Dr. Ingelfinger, Lynne McCusker, Dr. Gahl, Dr. Mannon, Dr. Kirk, Dr. Kleta, Susan Jordan and Tracy Cropper for working with him and continuing to make miracles happen in his life. Despite the challenges and adversity that cystinosis can bring, Shea has grown into a loving, happy, thriving young man.
A Tale of Two Transplants

Andrew Hoffman and Heidi Hughes at the NIH after Andrew’s transplant. Heidi was the first patient with cystinosis to receive a steroid-free kidney transplant at the NIH.

The Hammond Family—Colleen, Shea, Jack, and Kayla—six days after Colleen donated one of her kidneys to Shea at the NIH.

Andrew Hoffman and Tracy Cropper. Tracy is a Post Transplant Coordinator at the NIH.

Colleen and Shea Hammond spend time together prior to surgery. Colleen donated one of her kidneys to her son, Shea.
What the Cystinosis Research Network Means to Me

In celebration of its tenth birthday, the Cystinosis Research Network asked some families affected by cystinosis to comment on what CRN has meant to them. Here are some of the responses we received.

I would like to say that this group has been a great place to vent, cry and share advice and concerns. My daughter was diagnosed with Cystinosis in 2003. I visited the online support group for the first time with tears, thinking that no one could really know what was happening to my baby. I was overwhelmed when I opened up the web site and saw all the welcomes and embraces that were given to me over the computer. While we may all say that we have our share of troubles, there is always someone there offering help. We are all there like family. My family has always been there for us, but this group has helped deal with the frustration. In 2004, my newborn son was also diagnosed with Cystinosis, and I was heartbroken. I came to the group as a discouraged mama. When I sent the e-mail telling everyone about my son, again, I received heartfelt responses. I could feel everyone’s concerns and thoughts.

This group is a blessing to me because there is nothing that we have to face alone, and there is usually someone out there who has been there. If not, there is always someone willing to help with advice and encouragement. This group has been there for my family through it all.

Pam Smith
Mom to Gracie, 5 and Hunter, 3
both with cystinosis

CRN was the only place to really turn to after the kids were diagnosed. They had the real answers to the real problems in dealing with the day to day struggle of a very complex disease like cystinosis. It was such a comfort to become a part of a family that knew exactly what we were going through. Within a couple weeks of their diagnoses, we received phone calls from many CRN families and those who volunteer in the organization. We were also mailed myriads of information about the disease and the experts to contact. But by far, what has meant the most are the conferences that CRN funds. Meeting those afflicted and dealing with the disease just gave us so much hope. They had lives. They were thriving in many ways. We had to see it to believe it!

Brett and Brittney LeBeau
Parents to Alexis, 5 and Evan, 3
both with cystinosis

Evan and Alexis LeBeau
The CRN has provided for us a place to share our joys, fears, and frustrations concerning this disease. We have found a great family that knows what we have been through and shares all our emotions. The CRN has also provided us with great information that has helped us on our journey through this disease. We have not yet had the opportunity to attend a CRN conference but would love to in the future. They have in the past, before they started having their own conferences, provided us with scholarship money to attend a Foundation conference and have provided us with brochures for our Midwest Gathering. I would like to thank the CRN for all its support through the years and hope that they are around for a very long time.

The Gard family,
Eric, Dena, Nikolaus, without cystinosis, and Mikaela, 13, with cystinosis

CRN represents HOPE to us. Hope for a cure. Hope for a future where decisions in every day life don’t have to be weighed against medical consequences. Hope that someday our children will have the opportunity to sleep through the night, undisturbed by the regimented dosages of Cystagon.

Hope that it is recognized earlier in life. Hope for medical support who understands the implications of Cystinosis. Hope that the research done on Cystinosis not only helps our children but others who live with illness as well. Hope for the future for all of us whose lives have been touched by Cystinosis.

Leslie McGregor
Mom to Darcy, 22, without cystinosis, and Jared, 17, with cystinosis

I have two children with cystinosis. Jeffrey is 11 years old and Alicia is 7. The Cystinosis Research Network has touched my life by giving me back HOPE. When my kids were diagnosed, I felt helpless and alone. I felt like all of the hopes and dreams that I had for my beautiful children were gone---stolen by cystinosis. Then I discovered the CRN. It is so comforting to know that there is an entire network of people devoted to finding a cure for cystinosis. It is comforting to see the success of so many people who are living with cystinosis. With Hope and Faith and the CRN, anything is possible. Thank you.

Michelle Wachter
mom to Jeffrey 11 & Alicia 7 both with cystinosis and James, 2, without cystinosis

James, Jeffrey, and Alicia Wachter
What the Cystinosis Research Network Means to Me

CRN has given me HOPE. It has helped me provide accurate information, given me access to medical personnel who have treated this disease for years, given me a way to act – by volunteering, and connected me to a whole family that lives this life of cystinosis.

In November, 1996, at a routine 9 month check up for my son Garrett, the Physicians Assistant who was conducting the exam noted that he had stopped growing. We had already discussed the fact that he had no interest in baby food and only wanted to drink water and formula. Since this was my first child, I didn’t know that the amounts he was drinking were significant.

When she left the room to get his shots, she said; “He is probably just on the cusp of a growth spurt, you’ll see. But if he hasn’t grown but the next appointment, we’ll have to do a failure to thrive work up.”

My exhaustion from sitting up in a recliner nights with what other people were labeling my “fussy baby,” alternating between a water bottle and a formula bottle, turned to panic. When she returned to the room she had changed her mind, and decided to do the work-up right then. The PA knew me well enough to know I would worry for those three months now that those words were out in the open. The urine that she took that day did have blood, sugar and protein in it, and so we were sent off to the nearby hospital for some preliminary blood tests. When the Pediatrician called back, she said we would need to see a Pediatric Nephrologist (one of many new words Garrett’s father, Larry, and I would learn). Usually there was a two or three month lag before getting an appointment to see them. Based on the urine and blood results, they were going to see Garrett the next Wednesday, the day before Thanksgiving. Well, this made me worry even more.

At our first appointment at Children’s hospital in Buffalo NY, the Nephrologist told us that Garrett had Fanconi Syndrome, but that was secondary to the real problem. We needed to find out what was causing it. There were a couple of things it could be, one of which was cystinosis. Garrett got started on the supplements at that point in time. I started researching this cystinosis, and what I read was scary. Remember this was 10 years ago. Most of the information I read was outdated – and infants didn’t have the benefit of Cystagon.

At that point, our string of good luck ran out. A Czech medical fellow at the Kidney Center took over and getting the cystine test kit didn’t happen until after the New Year. The fellow spoke very broken English, and when he gave us the diagnosis over the phone (YES), we were told Garrett would die within a year. Of course, this corresponded with much of the research I had done.

Well, as Garrett likes to say – he has proven them wrong. He is 10, about to turn 11 in February. He has had a plethora of problems over the years but is active in inter-mural sports, water skis, rides his bike, and does many things I never anticipated those 10 years ago. The medical fellow moved on, and our medical team has been tremendous since then.
What the Cystinosis Research Network Means to Me

Hooking up with the CRN and becoming an active member of this group has been a life-changing choice. It has put me in control of this situation and reminds me constantly that I am his advocate.

- This group has actively moved forward in educating the medical community. There are so many metabolic diseases that medical students are many times given a <10 minute review of cystinosis to get them through the trick question on their exams. In fact, they are sometimes told they will never see a case. By going to medical conferences and having a booth, we show a commitment to building a relationship and networking within the medical community. On an individual basis, Garrett's hospital is a teaching hospital. Every visit we see a different resident, and so we try to share with them the basics – and tell them, “If you see cystinosis and diagnose it, you will probably save a life.”

- As a source of funding, CRN continues to choose medical studies that will benefit the most cystinosis patients. By putting together a Scientific Review Board (SRB), we have some of the finest resources in the world reviewing proposals and advising us on how to spend our hard earned dollars. Garrett has/is participating in these studies wherever it is possible/makes sense.

- By coming up with, running, and participating in fundraisers, I am helping out in raising money for everything we do—research, education and awareness, and family support. Family Support is probably the area of CRN in which I have been most active. This fits my personality and seems to come naturally to me. The fact that we have an online support group and a web site is so exciting compared to what there was 10 years ago. I know making a call to families that have been newly diagnosed is a giving thing for me. I listen to what they have gone through and tell them that it will get better. I tell them there are doctors who KNOW what is wrong and can speak with their doctor, and I let them know that we have family and medical conferences every other year where they can meet other families and people who live well with this disease.

CRN has come a long way in 10 years. We want to go much farther. Please do all that you can.

Lynn Thomas, Mom to Garrett, 10, with cystinosis
CRN Director – Family Support
Fundraising Brings us Closer to a Cure

Please consider hosting a fundraiser to help the CRN accomplish its ultimate mission of finding a cure for cystinosis. If you need more information, please contact the CRN at: CRN@cystinosis.org or 1 (866)-276-3669

Note Card Fundraiser

The CRN is selling lovely note cards to support cystinosis research. Give a gift that says you care and also supports a worthy cause.

Design options are floral, poinsettia, snowflake or a script initial. The inside is blank. The CRN logo and website address is on the back. A set of 8 cards and envelopes is $10.00 (US) which includes shipping and handling.

Go to http://www.cystinosis.org/news090804.html to download an order form.

Madison Feis Raffle in honor of Kellen Binger

By Jennie Smith

On June 3, 2006 in Madison, Wisconsin, the Trinity Academy of Irish Dance held an Irish Dance competition (feis) for 1600 dancers and their families. Judges and musicians came from Europe and the United States. This year the feis committee chose to donate the proceeds of our raffle to the Cystinosis Research Network in honor of Kellen Binger. Our Trinity community wanted to support the Binger family since Kellen had recently received a new kidney from his dad. Kellen’s older sister, Kole, has been dancing with Trinity for four years.

We knew we could count on Trinity families to make the raffle a success, but we were overwhelmed with the support from all the different Irish dance schools that attended. Our raffled raised $1316.00 for the CRN—more than double what the raffle has raised in the past!

We would like to thank the CRN for their input and ideas. It was so rewarding to do something positive to help out in the fight against cystinosis.

Kellen Binger stands next to his sister, Kole, and the Madison Trinity Irish dancers. Also included in the photo are Laura McNamara (Madison Trinity teacher), Natalie Sliwinski (Trinity Academy Director), and Ann McCarthy (Team Manager). The group raised $1316.00 for the CRN.
By Dawn Weiland

On August 26th, the third annual Family Fun Day in honor of Jack Greeley was held in Lincolnshire, Illinois.

Tamarak Country Day School and Camp was again the gracious host, providing the terrific venue and incredible help and support, as this year’s event was the biggest and best yet, despite clouds and sprinkles for part of the afternoon. Over 500 attendees enjoyed games, pony rides, great food, arts and crafts, face painting, tattoos, inflatable bounce house and football throw, sports, swimming and much more!

The raffle/silent auction proved very successful, as tremendous support from local businesses which donated items and services produced a profit of over $8,000.

We were also fortunate that two other families whose children have Cystinosis, the LeBeaus and the Julians, were able to attend Family Fun Day this year.

For the third year, the community truly embraced this event. We had over 50 volunteers manning the various stations, as well as donating their time, expertise and services to make the event run smoothly. It is the biggest event of its kind in Lincolnshire and wouldn’t exist without the support of this remarkable community.

In all, Family Fun Day raised nearly $18,000 this year, bringing its three year total to over $40,000. We are all so happy that Jack continues to do well, and are pleased that our efforts will help him and others through direct support of the Cystinosis Research Network.
3rd Annual Family Fun Day

Employees of West Bloomfield Township in West Bloomfield, Michigan raised $115 in honor of Jack Greeley. The funds were raised as a result of “Casual Friday” where each employee donated a minimum of $1.00 to attend work in casual clothing. Jack’s grandmother, Joyce Greeley, was a long time employee of West Bloomfield Township.
Carnival for a Cure in honor of The LeBeaus

By Brittney LeBeau

After many, many long months of planning our first fundraiser, Carnival for a Cure occurred and with great success. Held at Community Christian Church (CCC) in Naperville on Friday October 6th, we had over 350 people attend and raised over $18,000. There was a moon jump, slide, dance/dance revolution, food, games, face painting, pumpkin painting and much more for the youngest attendees as well as a silent auction, split the pot raffle and bake sale for the adults. The biggest success of the night was by far the Silent auction which brought in almost $7,000. Many items bid on were two pairs of Notre Dame tickets, Chicago Bears tickets, custom made furniture, jewelry, several vacation giveaways and many more.

We have many people to thank and who were apart of making this such a successful year. From local businesses to CCC, CRN, our volunteers and our Community. The Naperville Sun, our local newspaper, played a key role in advertising the Carnival for a Cure and getting the word out about Cystinosis and CRN. Other ways we got the word out were hanging posters in our schools, church, doctor's office, sending emails and simply word of mouth.

But none of this would have been possible if it weren't for our dearest friend Amber Stefanski! She guided the way and encouraged us to do this fundraiser from beginning to end. An Event Planner by vocation, she made many sacrifices with her time and money to make this come together. What a gift and blessing she has been to our family. We met her at church over 6 years ago when we were pregnant with Alexis, our first child. She carried us through our struggle to get a diagnosis for Alexis and eventually Evan. We had a shoulder to cry on, vent to, laugh with, and simply be real toward. No amount of vomiting, medications or hardships kept her from fulfilling her God-given purpose. A true Friend!
Sports Outings Make Great Fundraisers

A. Joseph DeNucci Golf Classic

Massachusetts State Auditor, Joseph DeNucci, has served as the Co-Chair of the Cystinosis Research Network Fundraisers in Newton, MA for many years. Joe and Russ Halloran (Shea Hammond’s grandfather) have teamed up to Chair the fundraisers to raise research funds to cure cystinosis. Joe's tremendous compassion and dedication are helping to restore the dreams of all who are affected by cystinosis. Special thanks to Joe and friends for designating the Cystinosis Research Network as the recipient of the funds raised from the A. Joseph DeNucci Golf Classic held in July which raised $5,530 for CRN. The total from the golf tournaments held over the years totals $19,140!

2nd Annual “Let’s Strike Out Cystinosis!” Bowling Fundraiser

This summer, Ina and Victor Gardner Sr. of Bedford Heights, Ohio were again able to host the second annual "Let's Strike Out Cystinosis" bowling event. "Let's Strike Out Cystinosis" was, according to all who attended, a resounding success. Approximately 300 people participated.

Word of the event was distributed by flyers and word of mouth and tickets were sold prior to the event and at the door. While a live DJ played, food that was donated by two local restaurants was served, and the participants were treated to a party atmosphere as they bowled. Desserts and other goodies were contributed by family and friends. A silent auction, with over 20 raffle prizes, also added to the festivities. The event raised approximately $3,000.00.

In addition, this year bumper bowling was added for the children and adults who might have needed it! It was noted with much amusement that Victor Jr. out-bowled his mom. His mom's comment to that was "As if getting beat by your 6 year old son is not bad enough 1 time..he is still asking to go bowling again!"

The Gardner's have two children—Victor, 7, and India, 4. Victor has cystinosis. The Gardners feel that the key to the success of the event was "their determination to give it their all and to remember all things are possible thru Christ. People do want to help and most people will support a good cause."

2nd Annual Golf Tournament in Honor of Nevaeh Stanford

Nevaeh Stanford was diagnosed with cystinosis at the age of 14 months. She is now 2 1/2 years old and doing great. The 2nd annual golf tournament held in her honor was on May 19, 2006 in her town. Nevaeh’s parents, Brandy and Jeremy Stanford, decided to donate $5000 from the tournament to the Cystinosis Research Network. They hope to do this each year to help not only their daughter but all individuals with cystinosis.

Nevaeh Stanford at the golf tournament in her honor on May 19, 2006
A Spooktacular Fundraiser!

Courtney Shepherd’s first Halloween party was more than just a party. She raised $440.00 for the Cystinosis Research Network! Courtney works with Brett LeBeau. When she met Brett and Brittney LeBeau’s children, Alexis and Evan, both with cystinosis, she knew she wanted to help in any way possible. Courtney had been involved in fundraising events in college and had been wanting to do fundraising again. She told her roommates she wanted to have a Halloween fundraising party for the CRN, and they agreed it was a worthwhile cause.

She sent out invitations and let guests know that it would be a fundraiser for cystinosis research. Courtney did not take money at the door but set up an optional donation jar. She had information about cystinosis, the CRN, and the LeBeau family near the jar as well as a picture of Alexis and Evan behind the jar. By the end of the evening, there was $440.00 in the jar for the CRN!

Courtney expressed that the CRN is an important charity for which to raise both money and awareness. Many people she talks to about cystinosis are saddened that it is so rare, and therefore, there is not very much funding for a cure or improved treatments. The Cystinosis Research Network appreciates Courtney’s efforts to raise money and awareness for cystinosis! Thank you, Courtney!

Are you ready for some MIRACLES?

Once again, Cystinosis Research Network will be hosting a dinner dance in Jericho, New York. The date of the dinner dance is Saturday, February 24, 2006 at the Millridge Cottage from 7-11 p.m., and the cost is $75 per ticket. Marybeth Krummenacker is the dinner Chair of this event and urges anybody who would like to attend to contact her at (516)931-6785 or by e-mail at MKrumm@aol.com. This event has proven to be a great night for both CRN and for those family and friends who have attended in the past. The last two events have raised a over $35,000 for CRN and have been earmarked for such events as the bi-annual family conference and various research projects. The 2007 event hopes to be the best ever. Anyone in the New York/Long Island area who might be interested in attending, please contact Marybeth. We have to make this year’s “Miracles at Milleridge” the best one yet!
Participants Sought for Cystinosis Research

The Pediatric Neurology Research Group at the University of California, San Diego (UCSD) is conducting the following research studies on cystinosis:

**Brain and Cognitive Development in Cystinosis**
The UCSD Pediatric Neurology Research Group is conducting a study to examine cognitive and brain development in individuals with cystinosis. Participants will include individuals with cystinosis ages 3-7 years and ages 12-17 years. This study will involve two 2-hour psychometric testing sessions that are scheduled at your convenience. The testing is non-invasive and behaviorally-oriented, and it helps us learn more about the cognitive and behavioral effects of cystinosis. Individuals will be asked to perform such tasks as putting together puzzles, defining words, and identifying or remembering pictures. Your participation in the psychometric testing is entirely voluntary.

As part of the study, Dr. Doris Trauner would like to conduct neurological examinations and Magnetic Resonance Imaging (MRI). Again, participation is voluntary. The neurological examination is a non-invasive exam that helps us learn more about an individual in terms of coordination, reflexes, sensory skills, language, and other neurologic functions. Individuals may be asked to do such tasks as kick a ball, stack blocks, and repeat sounds. MRIs are also considered safe and non-invasive and enable us to learn more about the structure of the brain.

Dr. Trauner hopes to examine any changes in the brain and cognitive development over time by seeing individuals for follow-up testing two years after their initial visit.

If you would like to participate in the Cognitive and Brain Development study or if you would like more information, please contact Jenny Williams at UCSD:

j7williams@ucsd.edu
858-822-6800

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**Executive Functioning in Cystinosis**
The UCSD Pediatric Neurology Research Group is conducting a study to examine executive functioning, which includes behaviors such as problem solving, planning, attention, and impulsivity, in individuals with cystinosis. Participants will include individuals with cystinosis ages 8 years through adulthood. This study will involve two 2-hour psychometric testing sessions. The testing is non-invasive and behaviorally-oriented, and it will help us learn more about executive functioning in cystinosis. Individuals will also be asked to fill out a questionnaire regarding behaviors such as planning and attention. Participation in this research study is completely voluntary.

If you would like to participate in the Executive Functioning study or if you would like more information, please contact Lynne Babchuck at UCSD:

lbabchuck@crl.ucsd.edu
858-822-6800

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Thank you very much for your time and consideration. We look forward to seeing you! We greatly appreciate the time and effort that families have devoted to increasing our knowledge of the effects of cystinosis.
Dr. Dohil, Dr. Schneider and Dr. Fidler at the University of California, San Diego, are looking for volunteers for their research study. They are currently looking to enroll patients for Spring 2007.

The aim of this research study is to find out if by making changes to the Cystagon tablet it will be possible to take Cystagon every 12 hours instead of every 6 hours. The changes made will stop the tablet dissolving in the stomach. It will instead dissolve in the small intestine. These changes are called enteric coating. The doctors believe that this way more Cystagon will get into the body and its positive effect will last longer. The substance being used for the coating is Eudragit and is produced by Röhm America in New Jersey.

The study will start in late spring 2006 and has 3 parts. During the first part the patient will continue their regular Cystagon therapy. The therapy will be monitored closely for 4 weeks by testing white blood cell cystine levels every week. These tests will be done at the patient’s regular physicians office.

After these 4 weeks, the patient will be flown to San Diego for some testing. The patient will have to take Cystagon in the morning of 3 different days, first regular Cystagon and then two different doses of coated Cystagon. After taking the tablets, a number of blood samples will be drawn during the next 12 hours. These blood samples will be used to measure the concentration of active component of Cystagon (cysteamine) in the blood as well as measure the white blood cell cystine levels. There will be one rest day between the study days, so the patient will be in the hospital for 5 whole days and 6 nights in total.

The last part of the study will again take place at home. The patient will be given coated Cystagon, which they have to take twice daily for 4 weeks. During this time the success of the therapy will be evaluated by measuring the white blood cell cystine levels every week. These tests will again be done at the patient’s regular physicians office. After 4 weeks the patient will resume taking regular Cystagon every 6 hours. The patient will not be able to continue the therapy with coated Cystagon.

For this study, the doctors are looking for about 12 volunteers with cystinosis. The volunteers have to be at least 6 years old, have so far not require a kidney transplant and have to be able to swallow tablets. The coated Cystagon tablets cannot work if they are opened and the contents is dissolved in water and because of this the patient has to always swallow the Cystagon tablets. Patient’s participation will also depend on whether their regular physicians can commit to seeing study patients.

Travel expenses for the volunteer and one parent to San Diego will be covered and the volunteers will be compensated for participating in this study. If you are interested in participating and would like more information, please contact Meredith Fidler, PhD, at (619) 543 2049 or mfidler@ucsd.edu.
Join The Cystinosis Research Network (CRN) and become part of a global network of caring families, concerned individuals and healthcare professionals working together in the fight against cystinosis. The Cystinosis Research Network’s vision is the discovery of improved treatments and ultimately a cure for cystinosis. The CRN is an all-volunteer; non-profit organization dedicated to sponsor and advocate research, provide family support, and educate the public and medical communities about cystinosis. The CRN funds research and programs primarily through donations from the public, grassroots fundraising events and grants.

CRN provides outreach and access to resources. We take great pride in carrying out our motto: “Searching For A Cure”...whether you are …

* A **Parent** who needs critical resource information, support services or help in sharing the challenges of cystinosis to those who serve your child.

* An **Adult** with cystinosis interested in information regarding medical and social issues that are specifically geared for adults.

* A **Relative** or a **Friend** who wants to increase their understanding of Cystinosis and find out how you can help out or become involved.

* A **Physician, Social Worker, Educator** or other **Professional** who makes a difference in the life of a family affected by cystinosis, and want to have access to critical information to better serve your patient, student or client.

Joining the Cystinosis Research Network enables you to:

* Receive all the latest Cystinosis information through our countless resources, including the CRN Newsletter, our very informative web page www.cystinosis.org, the ever popular online Cystinosis Support Group and our toll free number (1-866-276-3669).

* Attend the CRN Family Conference with other Cystinosis Families to exchange knowledge and create friendships. Also, find out the latest discoveries about cystinosis from the medical professionals, following their attendance of CRN’s Scientific Symposium, keeping current on scientific advances in Cystinosis research.

* Let your voice be heard by legislators and policymakers who need to know why Cystinosis (and other rare diseases) are important issues to you.

* Have access to the Cystinosis Research Network’s representatives in the areas that are most relevant at any given time to you or your loved one affected by Cystinosis.

**Join the Cystinosis Research Network today!**

Thank you for your consideration in becoming a member of the Cystinosis Research Network.

Pam Woodward
VP Family Support, Membership Committee, Chairperson
Cystinosis Research Network
Cystinosis Research Network Membership Form:

- Immediate Family: $20.00
- Extended Family / Friend: $25.00
- Professional: $35.00
- International: (Including Canada) Base rate (see above categories) plus $10.00 for postage. Payable in US dollars.

Please complete the form & mail with check payable to CRN to: CRN, 10 Pine Avenue, Burlington, MA 01803

Name______________________________________________________________
Street____________________________________________________________________
City & State__________________________ Zip Code__________ Country________
Phone__________________________ Fax_______________________Email_____________________________

Name of Child / Adult / Acquaintance / Patient affected with cystinosis: _________________________________

Please join a Cystinosis Research Network (CRN) Committee and make a difference in the quality of life of children, adults, and families affected by cystinosis. Many opportunities are available.

I am interested in joining a committee to help CRN. Please check: _______Yes ________No
If yes, please check the committee you would like to join or would like more information about:

- Family Support
- Development
- Research
- Finance
- Education / Awareness
- Membership
- Professional Advisory
- Other:

For more information, please call CRN at 1-866-276-3669, 781-229-6182 or email crn@cystinosis.org or visit www.cystinosis.org. Thank You!

Join A CRN Support Group

Looking for a way to communicate with others in the Cystinosis Community on a day-to-day basis?

The Cystinosis Research Network offers three email support groups for communicating with others in the cystinosis community:

The CRN Support Group is a group for parents, affected adults, caregivers, family, and friends. We also welcome researchers and medical professionals who are interested in cystinosis. This is the place to discuss the various aspects of cystinosis, and how it affects our lives, how we cope, vent our frustrations, share our fears, our hopes, and our dreams.

The CRN Teen Support Group is for teens with cystinosis and teenage siblings of children and adults with cystinosis. Connect with other teenagers who are dealing with similar issues. The posts include questions, concerns, ideas and supportive sharing.

The CRN News Group is for all who would like to receive announcements of upcoming events, important activities and breakthroughs.
Please Help CRN In Our Mission of Finding A Cure and Improved Treatments, Providing Family Support, and Raising Awareness

☐ YES, I want to help children and adults with cystinosis.

Enclosed is my tax deductible contribution of: $_________ made payable to the Cystinosis Research Network (CRN) and mail to: 10 Pine Avenue, Burlington, MA 01803

Name___________________________________________________________

Street________________________________________________________________________

City & State_______________________ Zip Code________________________

Phone________________ Fax________________ Email______________________

In Honor Of__________________________________________________________

In Memory Of________________________________________________________

You may send notification of my gift to:

____________________________________________________________________________

United Way and You

The United Way begins its annual fundraising drive each fall in nearly every community in the United States. It provides a way for many charities to obtain funds by individuals contributing a small portion of their paycheck through payroll deduction. Each United Way chapter lists a variety of organizations that may be chosen for payroll deductions, most of which are locally based. The majority of United Way chapters allow individuals to write in the charity of their choice. Even though we are a national organization, our 501(c)(3) nonprofit status qualifies us for this write-in option. CRN’s Federal Tax ID # is: 04-3323789.

To initiate a write-in campaign in your workplace, contact your company’s United Way chairperson to ask if there is a write-in option and if so, the possibility of informing the rest of the employees about the special needs of cystinosis research. Most coworkers are eager to donate to a cause that is personally tied to a colleague. Posting a public letter or a poster asking your fellow employees to join you in our fight against cystinosis might be one way to reach all the employees and increase our funding. If you or your United Way chairperson has any questions, please call CRN at toll free: 1-866-276-3669 or 781-229-6182.

Donation Reminders

- Don’t forget you can donate online through Network for Good. Visit www.cystinosis.org for more information.

- Don’t forget about your company’s matching-gift programs.

Any gift, large or small, is greatly appreciated!
The Cystinosis Research Network expresses its gratitude for donations, memorial gifts, and “in your honor” gifts. Thanks to all for giving a gift that continues to give. With the financial support of members and friends, the Cystinosis Research Network continues our mission of supporting cystinosis research, cystinosis education programs, and providing family support. We wish to thank the following individuals and groups whose donations were received between May 23, 2006 and October 26, 2006.

**Gifts and Donations**

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<th>Leadership Circle ($5,000 - $15,000)</th>
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<td>Sigma Tau Pharmaceuticals</td>
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<th>Ambassadors ($2,500 - $4,999)</th>
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<td>A. Joseph DeNucci Golf Tournament in honor of Shea Hammond</td>
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<td>Dahn Maier through United Way of New York City in honor of Christian Morales</td>
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<td>Velyna Morales through United Way of New York City in honor of Christian Morales</td>
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<td>Amy Shelton McNutt Charitable Trust in honor of John Ben and Ava Shepperd</td>
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<td>Brandy and Jeremy Stanford &amp; Friends Golf Tournament in honor of Neveah Stanford</td>
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<td>Tamarack Family Fun Day Cash Donations in honor of Jack Greeley</td>
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<td>Sharen Jester Turney through United Way of New York City in honor of Christian Morales</td>
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<th>Benefactors ($1,000 - $2,499)</th>
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<td>Larry and Pat LeBeau-Carnival for a Cure in honor of Alexis and Evan LeBeau</td>
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<td>Mark and Denice Flerchinger in honor of Tina Flerchinger</td>
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<td>Ina Gardener Fundraiser in Honor of Victor Gardener, Jr.</td>
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<td>Madison Feis Committee - Trinity Booster Club in Honor of Kellen Binger</td>
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<td>David Pullen in honor of Mitchell Smith</td>
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<td>Sam's in honor of Jack Greeley</td>
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<td>Shirley Schindler in honor of Heidi Hughes</td>
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<td>Baystate Properties Real Estate – DeNucci Golf Tournament in honor of Shea Hammond</td>
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<td>William and Cicily Brogan Fund in honor of Jack Greeley</td>
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<td>Jamie Goldberg in honor of Jack Greeley</td>
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<td>David, Diane, and Katherine Hankle in honor of Garry, Carol, Jennell and Heidi Hughes</td>
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<td>Susanne Immell in honor of Jack Greeley</td>
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<td>Jim and Beth Konieczny in honor of Jack Greeley</td>
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<td>Sam Bloodgood in honor of Jack Greeley</td>
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<td>Christopher Burke LLC, John and Marianne Wills- Carnival for a Cure in honor of Alexis and Evan LeBeau</td>
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<td>Yvonne and Wallace Dvorak in honor of Jack Greeley</td>
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<td>Michelle Crotty- Carnival for a Cure in honor of Alexis and Evan LeBeau</td>
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<td>Darci Drury- Carnival for a Cure in honor of Alexis and Evan LeBeau</td>
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<td>Nick and Julie Giannasi- Carnival for a Cure in honor of Alexis and Evan LeBeau</td>
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<td>Kathy Gordon in honor of Jack Greeley</td>
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<td>David and Joy Holecek in honor of Jack Greeley</td>
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<td>Cheryl -Anne and Daniel Howell - DeNucci Golf Tournament -in honor of Shea Hammond</td>
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<td>Michelle Kaplan in honor of Jack Greeley</td>
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<td>Les and Connie Larson- Carnival for a Cure in honor of Alexis and Evan LeBeau</td>
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<td>Linda and Michael Leone - DeNucci Golf Tournament - in honor of Shea Hammond</td>
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<td>Mark and Mindy Leonard in honor of Jack Greeley</td>
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<td>Daniel and Leigh Martin- Carnival for a Cure in honor of Alexis and Evan LeBeau</td>
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<td>Mike and Rae Mattson- Carnival for a Cure in honor of Alexis and Evan LeBeau</td>
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<td>Eric and Erin Melby- Carnival for a Cure in honor of Alexis and Evan LeBeau</td>
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<td>Rosalind Moore in honor of Victor Gardener, Jr.</td>
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<td>Michael and Julie Muth in honor of Jack Greeley</td>
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<td>Panson Electrical Services</td>
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<td>John and Susan Ruddy- Carnival for a Cure in honor of Alexis and Evan LeBeau</td>
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<td>Dan and Theresa Schindler- Carnival for a Cure in honor of Alexis and Evan LeBeau</td>
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<td>Paul and Laura Russo in honor of Jack Greeley</td>
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<tr>
<td>Elva Smith in commemoration of the wedding of Antonya Jandacek and Ryan Sanders and in honor of Mitchell Smith</td>
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<td>Thomas Terrio - DeNucci Golf Tournament - in honor of Shea Hammond</td>
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<tr>
<td>John and Pucky Zimmerman- Carnival for a Cure in honor of Alexis and Evan LeBeau</td>
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Supporters ($50 - $249)

David and Joan Allemeier- Carnival for a Cure in honor of Alexis and Evan LeBeau
Susan Applegate- Carnival for a Cure in honor of Alexis and Evan LeBeau
Chad and Michelle Ashby- Carnival for a Cure in honor of Alexis and Evan LeBeau
Mike and Elice Altamore- Carnival for a Cure in honor of Alexis and Evan LeBeau
Douglas and Lisa Ayers in honor of Alex Weaver
Georgene Bell in honor of Louise Limacher
Bethany Christian Church in honor of Victor Gardener, Jr.
Syed and Nooresha Biabani- Carnival for a Cure in honor of Alexis and Evan LeBeau
Armen and Judy Biberian- Carnival for a Cure in honor of Alexis and Evan LeBeau
Thomas Bondurant in honor of Alex Weaver
Spencer and Brandi Campbell- Carnival for a Cure in honor of Alexis and Evan LeBeau
Campbell and Campbell, PC in honor of Alex Weaver
CDG of Broomfield in honor of Neveah Standford
Christine Casale- Carnival for a Cure in honor of Alexis and Evan LeBeau
Duane and Kelley Chudy in honor of Jack Greeley
John and Amy Click in honor of Alex Weaver
Sarah Cohen in honor of Jack Greeley
Thomas Concannon from DeNucci Golf Tournament in honor of Shea Hammond
Paul and Idele Crimmins in honor of Jack Greeley
Dacono Properties, LLC in honor of Neveah Standford
Gerald and Elaine Daley in honor of Heidi Hughes
Shad Reed and Dalhart High School Basketball Team in honor of Mason Reed
Stephen Dalton in honor of Alex Weaver
Keith Degen- Carnival for a Cure in honor of Alexis and Evan LeBeau
Richard and Annette Ellspermann- Carnival for a Cure in honor of Alexis and Evan LeBeau
Michael and Victoria English- Carnival for a Cure in honor of Alexis and Evan LeBeau
J. Michael Estes in honor of Alex Weaver
Mike and Kim Fanella- Carnival for a Cure in honor of Alexis and Evan LeBeau
Harry and Patricia Field- Carnival for a Cure in honor of Alexis and Evan LeBeau
Bernard and Rose Finn in honor of Janice Finn
Karen Francis-Carnival for a Cure in honor of Alexis and Evan LeBeau
Linda and John Frye in honor of Alex Weaver
Robert and Gretchen Funck in honor of Jack Greeley
Gary and Kathryn Gordon in honor of Jack Greeley
Joshua and Lisa Gordon in honor of Jack Greeley
Charles and Joyce Greeley in honor of Jack Greeley
David and Amelia Hendryz- Carnival for a Cure in honor of Alexis and Evan LeBeau
Karen Homer- Carnival for a Cure in honor of Alexis and Evan LeBeau
Harrison Hubbard in honor of Alex Weaver
Hixon MFG and Supply Co. in honor of Neveah Standford
Hutchens and Hutchens, PC in honor of Alex Weaver
Patricia Jette in honor of Jack Greeley
Carl and Brenda Johnson in honor of Alex Weaver
Patricia Jones- Carnival for a Cure in honor of Alexis and Evan LeBeau
Lee and Barbara Joslin- Carnival for a Cure in honor of Alexis and Evan LeBeau
Michael Kelly in honor of Alex Weaver
Kraft Foods Matching Gift of Gregory Immell
Stephen Laulettta in honor of Jack Greeley
Gerard and Colleen Leider in honor of Jack Greeley
Susan Lewis- Carnival for a Cure in honor of Alexis and Evan LeBeau
Lincolnshire Academy of Dance in honor of Jack Greeley
Lincolnshire Rotary Charitable Fund in honor of Jack Greeley
LJD Enterprises, INC in honor of Neveah Stanford
Bob and Linda Mangum in memory of Jennifer Danielle Scharf
Keith Marcus in honor of Alex Weaver
Marlboro High School Class of 2000 in memory of Mark Stetson
Steven Marks in honor of Alex Weaver
Christopher and Angie Martin- Carnival for a Cure in honor of Alexis and Evan LeBeau
Elaine and George Martin in honor of Alex Weaver
Scott and Leslie Massow- Carnival for a Cure in honor of Alexis and Evan LeBeau
Mattel Children's Foundation matching gift to Paula Shal's donation
John McGarvey in honor of Alex Weaver
Tim McKeown and Mary Resing in honor of Jack Greeley
Anna McWilliams in honor of Victor Gardener, Jr.
Wayne Morgan in honor of Alex Weaver
Ned Mikula in honor of Alex Weaver
Doris Muller in honor of Janice Finn's birthday
Virginia Munoz in honor of Alex Weaver
National Football League matching gift of Nancy Behar and Ana Pettiti in honor of Jack Greeley
Patricia Jones Parker through United Way of Piedmont in honor of Laura McGinnis
Sarah and Teddy Parker in memory of Mark Stetson
Toni Parrish-- Carnival for a Cure in honor of Alexis and Evan LeBeau
Karla Pearson- Carnival for a Cure in honor of Alexis and Evan LeBeau
Mark and Jalaine Peterson in honor of Jack Greeley
Norma and James Petet in memory of Nancy Behar and Ana Petitti in honor of Jack Greeley
Jennifer Danielle Scharf
Wayne Morgan in honor of Alex Weaver
Thomas Concannon from DeNucci Golf Tournament in honor of Shea Hammond
Paul and Idele Crimmins in honor of Jack Greeley
Loretta Ross in honor of Alex Weaver
Suzanne Sackmann- Carnival for a Cure in honor of Alexis and Evan LeBeau
Heather Salazar in honor of Jack Greeley
Cheryl Sands in honor of Jack Greeley
Mark and Jennifer Sansone- Carnival for a Cure in honor of Alexis and Evan LeBeau
Loren and Kristine Schiro- Carnival for a Cure in honor of Alexis and Evan LeBeau
Peter Shamburek- Carnival for a Cure in honor of Alexis and Evan LeBeau
Elva Smith in honor of Mitchell Smith and in memory of Emanuel Livaudais, Jr.
Specified Tile and Flood Covering in honor of Lee Weaver and in honor of Alex Weaver
Anton Stelly in honor of Alex Weaver
Linda Sterling- Carnival for a Cure in honor of Alexis and Evan LeBeau
Hershel and Wanda Chenault in honor of Alex Weaver
James and Monica Fairman in honor of Jack Greeley
Mme. Elva Smith in honor of Mitchell Smith and in memory of Lee Weaver in honor of Alex Weaver
Christina K. Shepperd in honor of Jack Greeley

Contributors ($50 - $99)

Ali Amrishah in honor of Alex Weaver
Shirley Bartlett in memory of Margaret Hammond
Norman and Judi Beauregard- Carnival for a Cure in honor of Alexis and Evan LeBeau
Richard and Leigh Benowitz in honor of Shea Hammond
Steven and Cynthia Bensen in honor of Megan Morrill
Jason and Kari Berg- Carnival for a Cure in honor of Alexis and Evan LeBeau
Brown, Martin, PC in honor of Alex Weaver
Claire and Gregory Carr in honor of Alex Weaver
Paul and Michelle Cavzos- Carnival for a Cure in honor of Alexis and Evan LeBeau
Ramon Chalkley in honor of Alex Weaver
Hershel and Wanda Chenuault in honor of Victor Gardener, Jr.
Thomas Collins in honor of Alex Weaver
Carolyn Conwell in honor of Victor Gardener, Jr.
Montgomery and Colleen Cornell in honor of Jack Greeley
Robert and Lori DeMyer- Carnival for a Cure in honor of Alexis and Evan LeBeau
Josephine DiCesare- Carnival for a Cure in honor of Alexis and Evan LeBeau
William J. Doran III, J.D. in honor of Alex Weaver
Mary Jo Dunn- Carnival for a Cure in honor of Alexis and Evan LeBeau
James and Monica Fairman in honor of Jack Greeley
Richard Fuller in honor of Alex Weaver
Ron and Kandi Gasperini- Carnival for a Cure in honor of Alexis and Evan LeBeau
Robert Geary in honor of Alex Weaver
Sylvia Geerdink
Get it Straight Orthodontics in honor of Jack Greeley
David and Christy Greeley in honor of Jack Greeley
David and Christy Greeley- Carnival for a Cure in honor of Alexis and Evan LeBeau
Russ and Margaret Halloran in memory of Mark Stetson
Richard and Beverly Halter in memory of John Ben and Ava Shepperd

Rev. Robin Hedgeman in honor of Victor Gardener, Jr.
Harry and Cynthia Hutcherson in memory of Lee Weaver and in honor of Alex Weaver
Greg and Susanne Immell in honor of Jack Greeley
Rod and Laurie Jones in memory of Mr. Shurley Burnett
Ronald and Carol Kaplan in honor of Jack Greeley
D and K Keevins in honor of Jack Greeley
James Kessel in honor of Alex Weaver
Christopher and Debra Kiah in honor of Jack Greeley
Wade Kizer in honor of Alex Weaver
James and Elizabeth Konieczny in honor of Jack Greeley
Marvin and Roberta Kloptad in memory of Bernice Methe, Melvin Suko and Stanley Wrotny and in honor of Raymond Moreno, Jr.
Michael and Lara Kral- Carnival for a Cure in honor of Alexis and Evan LeBeau
Leonard Lambert in honor of Alex Weaver
Barbara and Gerald Larson in honor of Jack Greeley
Wally and LouAnn LeBeau- Carnival for a Cure in honor of Alexis and Evan LeBeau
Michael and Michelle MacLain- Carnival for a Cure in honor of Alexis and Evan LeBeau
Janice and David Mann in honor of Jill Morrill's birthday and Megan Morrill
Mildred McFlela in honor of Britney Carroll's Birthday
Patrick and Nancy O'Connell- Carnival for a Cure in honor of Alexis and Evan LeBeau
Robert and Sarah Parkerson in honor of Alex Weaver
John Parsons in honor of Alex Weaver
Kris and Elizabeth Patras- Carnival for a Cure in honor of Alexis and Evan LeBeau
Jodi Pauly in honor of Jack Greeley
Robert Petranek- Carnival for a Cure in honor of Alexis and Evan LeBeau
Debra Ponce de Leon in honor of Jack Greeley
Raul Prado in honor of Christian Morales
Chris and Jackie Price- Carnival for a Cure in honor of Alexis and Evan LeBeau
Dr. and Mrs. Ralston in honor of Corbin Rico and Helen and Bill Morable
Duncan Reid in honor of Alex Weaver
Larry Reinert in memory of Mr. Shurley Burnet
Lisa and Mark Reposa in memory of Mark Stetson and in honor of Shea Hammond
Joel and Marcia Resnick in honor of Jack Greeley
Ruhl Landscaping, Jim Ruhl- Carnival for a Cure in honor of Alexis and Evan LeBeau
Paul and Laura Russo in honor of Jack Greeley
Paul and Amy Atwater in honor of Jill Morrill’s birthday and Megan Morrill
Iris Seijo in honor of Janice Finn
John and Maureen Skei in honor of Janice Finn
Chris and Molly Simpson- Carnival for a Cure in honor of Alexis and Evan LeBeau
Stan and Bette Slazas-Carnival for a Cure in honor of Alexis and Evan LeBeau
David and Kristin Sliwicki in honor of Jack Greeley
Tondrowski and Associates PC in honor of Alex Weaver
Jeanne and Franklin Top in honor of Jack Greeley
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Maarten and Anne Van Gerven in honor of Jack Greeley
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Peggy and Ed Wright and Family in memory of Mark Stetson

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Dean and Janet Atterberry in memory of Mary Louise Schmidt
Chris and Amy Atwater in honor of Jill Morrill’s birthday and Megan Morrill
Jerry and Sheryl Bach in honor of Alex Weaver
Rob and Bridget Bachara- Carnival for a Cure in honor of Alexis and Evan LeBeau
Dr. Paul Baird in memory of Sierra Woodward
Nancy Banning in memory of Mr. Shurley Burnet
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Brian and Daphne Berryhill in honor of Jack Greeley
Joseph and Yvette Blank in honor of Jack Greeley
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Richard and Elizabeth Brandt in honor of Jack Greeley
Ronald and Janie Braun- Carnival for a Cure in honor of Alexis and Evan LeBeau
Irving and Esther Braunstein in honor of Glenn Jones
Jamie Bressner- Carnival for a Cure in honor of Alexis and Evan LeBeau
James and Julie Bridgeman- Carnival for a Cure in honor of Alexis and Evan LeBeau
Janet Brown in honor of Alex Weaver
Tanetta Brown in honor of Alex Weaver
Jeff and Tammy Bruner- Carnival for a Cure in honor of Alexis and Evan LeBeau
Martha and William Buchan in honor of Jack Greeley

Chris and Sharon Burnham- Carnival for a Cure in honor of Alexis and Evan LeBeau
Stanley and Ilene Burmood in memory of Sierra Woodward
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Wendy Condon- Carnival for a Cure in honor of Alexis and Evan LeBeau
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Audrey Dallamora in memory of Mark Stetson
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Carol Demson in memory of Sierra Woodward
Jeffrey and Angela Depew in honor of Jack Greeley
Jeffrey and Aimee Devlin in honor of Jack Greeley
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John and Meredith Driscoll in honor of Jack Greeley
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Dwight and Robin Ekenberg in honor of Jack Greeley
Marcia Elkin in honor of Jack Greeley and Lisa Endrud: Carnival for a Cure in honor of Alexis and Evan LeBeau
Thomas and Debra Eubank in honor of Alex Weaver
Saadia and Jean-Aicard Fabien in honor of Jack Greeley
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Arthur Filete in honor of Laura Krummenacker
Guy and Annette Fillett: Carnival for a Cure in honor of Alexis and Evan LeBeau
Michael and Debra Fisch in honor of Jack Greeley
Catherine Flerchinger in honor of Tina Flerchinger - Lemonade Stand
Monica Flynn in memory of Mark Stetson
Kay Frye in honor of Alex Weaver
Steven and Lynn Gackenbach in honor of Jack Greeley
Paul and Anne Galanides in honor of Alex Weaver
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Dovetta Garland in honor of Victor Gardener, Jr.
Bill and Jenny Garrison: Carnival for a Cure in honor of Alexis and Evan LeBeau
Judy Garrison: Carnival for a Cure in honor of Alexis and Evan LeBeau
Charles and Heather Gavin in honor of Alex Weaver
Craig and Cathy Gentry: Carnival for a Cure in honor of Alexis and Evan LeBeau
Vivian Gillings in honor of Victor Gardener, Jr.
Diane Gillis in honor of Jack Greeley
David Giroux in honor of Alex Weaver
Harriet and Barry Goldberg in honor of Jack Greeley
Thomas and Jean Gontkovic in honor of Alex Weaver
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Steve Hart: Carnival for a Cure in honor of Alexis and Evan LeBeau
Susan Hawkingson: West Bloomfield Township in honor of Jack and Joyce Greeley - Casual Friday Fundraiser
Gregory and Robyn Hebard in honor of Jack Greeley
Pat Helson: West Bloomfield Township in honor of Jack and Joyce Greeley - Casual Friday Fundraiser
Harry and Maggie Hemphill in honor of Victor Gardener, Jr.
Philip and Marlene Herbert: Carnival for a Cure in honor of Alexis and Evan LeBeau
Michelle Herron: Carnival for a Cure in honor of Alexis and Evan LeBeau
Michael and Wendy Herst in honor of Jack Greeley
June Holber in honor of Jack Greeley
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Dean Holden: Carnival for a Cure in honor of Alexis and Evan LeBeau
Jeffrey Hooper in honor of Jack Greeley
Dave and Stephanie Hulthen: Carnival for a Cure in honor of Alexis and Evan LeBeau
Keith Hurley in honor of Alex Weaver
Daniel and Mary Hylek: Carnival for a Cure in honor of Alexis and Evan LeBeau
Carl and Brenda Johnson in honor of Alex Weaver
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Michelle Kramer in honor of Jack Greeley
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Michelle McCarthy Dawson in honor of Alex Weaver  
Leslie McGregor in memory of Lindsay Wells and in honor Of Jared McGregor  
MG McKenna in memory of Mark Stetson  
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Troy and Janet McMahon- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Cary and Rande McMillan in honor of Jack Greeley  
Kent and Kirsten Melum- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Erin and Eric Metcalf- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Glenn Miller in memory of Mr. Shurley Burnett  
Jeff and Janet Miller- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Nate and Jean Miller- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Silvano and Christina Minella in honor of Skyler Minella  
Audrey Mital- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Kathleen Ann Mueller in honor of Alex Weaver  
David and Kathy Murray- Carnival for a Cure in honor of Alexis and Evan LeBeau  
William Nash in honor of Victor Gardener, Jr.  
Debra and Mark Nedervelt in honor of Alex Weaver  
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Steven and Jennifer Nenn- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Jill and Chris Newton in honor of Jack Greeley  
Stacie Nikkel- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Bryan and Amy Nottke- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Carol and Kenneth Oates in memory of Mark Stetson  
Ken Olsen- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Dan and Joan Patten in honor of Alex Weaver  
Isabel Paul in honor of Alex Weaver  
Patricia Pawelka in honor of Jill Morrill’s birthday and Megan Morrill  
Martha Perez- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Nancy and Michael Perry in memory of Mark Stetson  
Richard and Stephanie Pither in honor of Jack Greeley  
John and Kathy Paolello- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Bob Proctor- Carnival for a Cure in honor of Alexis and Evan LeBeau  
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Christopher and Jan Rabin in honor of Jack Greeley  
Dave and Dana Rank- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Lawrence and Ann Reilly in honor of Jack Greeley  
N. Richmond in honor of Alex Weaver  
Laura Ricks- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Pam and Bill Ricks- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Brian Robison in memory of Leamon C Weaver and in honor of Alex Weaver  
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Martin and Heila Rowan- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Anne and Timothy Ryan in honor of Jack Greeley  
Betty Sacra in honor of Alex Weaver  
Eugene Schmidt in memory of Mary Louise Schmidt  
Albina Schmidt in memory of Mary Louise Schmidt  
Marilyn Schumacher in memory of Mary Louise Schmidt  
Daniel and Theresa Sears -West Bloomfield Township in honor of Jack and Joyce Greeley - Casual Friday Fundraiser  
Amy Shapiro in honor of Jack Greeley  
Michael and Molly Shapiro in honor of Jack Greeley  
Joan Sharp- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Jim Shirley- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Chad and Lisa Simon- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Sandra and Douglas Simon in honor of Jack Greeley  
Brian and Erin Smith- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Dennis and Sheri Lynn Smith- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Mickey and Laura Smith- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Sheri Smith- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Yvonne and Wallace Smith in honor of Alex Weaver  
Norma Snively in memory of Mary Louise Schmidt  
Rudy and Mary Lou Stefanski- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Dana and Lee Stein in honor of Jack Greeley  
Tami and Scott Stensby in honor of Jack Greeley  
Charles Stevens in memory of Lee Weaver  
Becky Stines- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Taylor Stone in honor of Alex Weaver  
Todd B. Stone in honor of Alex Weaver  
Mary Stuart- Carnival for a Cure in honor of Alexis and Evan LeBeau  
Jennifer and James Sturgeon in honor of Jack Greeley
Layne and Starlyn Summers- Carnival for a Cure in honor of Alexis and Evan LeBeau
Anne Susina- Carnival for a Cure in honor of Alexis and Evan LeBeau
Joseph and Antoinette Talanges in honor of Jack Greeley
Lucia and David Thoensen in honor of Jack Greeley
James and Nancy Thomas in honor of Jack Greeley
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Scott and Elizabeth Treiber in honor of Jack Greeley
Christine Udoni in honor of Jack Greeley
Patrick and Julie Uplegger- Carnival for a Cure in honor of Alexis and Evan LeBeau
Carmine and Meghan Vozzolo- Carnival for a Cure in honor of Alexis and Evan LeBeau
Richard and Ann Wachter in honor of Jack Greeley
Susan Walsh- Carnival for a Cure in honor of Alexis and Evan LeBeau
Paul and Katy Warner- Carnival for a Cure in honor of Alexis and Evan LeBeau
J Wasik in honor of Victor Gardener, Jr.
Steve and Gina Wehde- Carnival for a Cure in honor of Alexis and Evan LeBeau
Dawn Weiland in honor of Jack Greeley
Michael and Randi Weis in honor of Jack Greeley
Judy Wells in memory of Lindsay Wells
Sari Wiaz in honor of Jack Greeley
Candace Wickstrom in memory of Sierra Woodward
Charles and L. Ann Wilkinson in memory of Sierra Woodward
Kim Williams- Carnival for a Cure in honor of Alexis and Evan LeBeau
Reinhard and Linda Williams in honor of Alex Weaver
Esther Windmueller in honor of Alex Weaver
Leslie Winton in honor of Jack Greeley
Charles and Beckie Wooster in memory of Sierra Woodward
Kerry and Andrea Wortzel in honor of Alex Weaver
Pam Wright- Carnival for a Cure in honor of Alexis and Evan LeBeau
Troy and Audrey Wright in memory of Mary Louise Schmidt
Meredith and Robert Yolles in honor of Jack Greeley

In Kind Donation
Tim Miller and TH Robison Inc., Furniture Donation in memory of Sierra Woodward
Vision and Mission Statement

**Vision.** The Cystinosis Research Network’s vision is the discovery of improved treatments and ultimately a cure for cystinosis.

**Mission.** The Cystinosis Research Network is a volunteer, non-profit organization dedicated to supporting and advocating research, providing family assistance and educating the public and medical communities about cystinosis.

Mark Your Calendars for Upcoming Conferences!

**July 19-21, 2007**
The Cystinosis Research Network Family Conference at the Hotel Contessa in San Antonio, Texas. More information will be released soon at www.cystinosis.org

**August, 2010**
International Pediatric Nephrology Meeting in New York City. More information will be on www.cystinosis.org as the event gets closer

When moving, please remember to notify the Cystinosis Research Network of your new address. We do appreciate it!