Rare Disease Advocate Cindy Abbott to Kick off 2011 Family Conference

Reaching Beyond the Clouds:
From Undiagnosed to Climbing Mt. Everest

The Cystinosis Research Network is pleased to announce that Cindy Abbott will kick off the 2011 CRN Family Conference in San Francisco as our Inspirational Speaker on Friday, July 15th. On August 1st 2007, she was diagnosed with Wegener’s Granulomatosis and on May 23rd 2010, she summited Mt. Everest. With a rare and potentially life-threatening disease, at the age 51 and functionally blind in one eye she became one of the 400 Americans who have climbed the world’s tallest mountain. Her journey to Mt. Everest began three years ago but her story, like so many other victims of rare unknown diseases, started more than a decade earlier. Please go to Cindy's website for more information [www.reachingbeyondtheclouds.com](http://www.reachingbeyondtheclouds.com).

See below for more on her amazing story:

"On August 1st 2007, I was diagnosed with Wegener’s Granulomatosis and on May 23rd 2010, I summited Mt. Everest. With a rare and potentially life-threatening disease, at the age 51, and functionally blind in one eye: I joined an elite group of American females who have climbed the world’s tallest mountain. My journey to Mt. Everest began three years ago but my story, like so many other victims of rare unknown diseases, started more than a decade earlier. At the age of 48 and with no mountaineering experience, I decided to climb Mt. Everest. A few months later I lost most of my vision in one eye and was diagnosed with a very rare potentially fatal disease, but I would not give up my dream to climb Everest. After 2.5 years of training, medical set-backs, and personal sacrifice; I became one of the 157 non-Sherpa people to summit the South Side of Mt. Everest in 2010.

About 3000 individuals have summited Mt. Everest and of those only 250 were female. Only about 400 Americans have summited Everest and of those about 40 were female. There are 30 million Americans with rare diseases and on international level there is no scale to measure to numbers affected. Before my climb I became an advocate to raise rare disease awareness for the National Organization of Rare Disorders (NORD) and the Vasculitis Foundation (VF). After 54 days of working my way up the mountain, I stood at the top of the world holding a NORD Banner."

Please plan to join us in San Francisco for the Family Conference, July 14-16 at the Hotel Nikko! Registration materials, information on conference scholarships and the conference agenda are available on the CRN website and have been mailed to families. Early bird registration ends in June and hotel room availability cannot be guaranteed after that time. Please feel free to contact CRN with any questions at [info@cystinosis.org](mailto:info@cystinosis.org).
Message from the President

Christy Greeley

By Christy Greeley

I have to be honest with all of you, I was really challenged in writing this newsletter’s article. I reached out to a friend to get their perspective. I was at a loss, not wanting to just summarize what is available in this newsletter or repeat what was included in the last edition. I really wanted the article and messages to be enlightening and encouraging. My friend suggested that I think about communicating what keeps me involved and committed to CRN and our cystinosis community 10 years after my son’s diagnosis. So, what and who inspires me?

Some of the sources of inspiration are more obvious than others: Individuals such as Bill Gahl, Jess Thoene and Jerry Schneider who are the pioneers in the diagnosis and treatment of cystinosis; specific physicians such as CRN’s NIH Cystinosis Fellow Galina Nesterova who has committed three years of her career to focus on studying and treating this disease to become a clinical expert in the disorder; physicians who serve on CRN’s Medical Advisory Committee and Scientific Review Board, who unselfishly devote so many hours to assisting our community in a variety of ways. There is little fame and fortune to be had by championing a rare disease. We owe our children’s lives to these individuals and they have my undying respect and gratitude.

But there are numerous “unsung heroes” who contribute so much to CRN and the entire community that are less obvious. Currently 24 individuals serve on CRN’s Board of Directors – 17 general members and 7 Executive Committee members. All live with cystinosis or care for their children or grandchildren living each day with it. All have made significant contributions. Let me tell you about a few.

Mack Maxwell is one of the most positive people I know. He lives his life to the fullest, traveling around the world and keeping up with his career, managing his cystinosis with grace. He also single handedly manages the CRN website, so efficiently that sometimes changes are made only minutes after the request is sent out – I don’t know how we would function without him.

When I think of Paula Shal, I think of unselfishness. She has a successful career, a wonderful and supportive family and still finds time to take care of one of the most time consuming job at CRN, Newsletter Editor. In fact, the night before her last transplant she was up until 2am trying to finish an edition of the newsletter so she would not leave too much work for others. Incredible dedication to all of us.

Elva and Lorna Smith are two of the bravest women I know. At the depths of their grief, only days after losing a son and a husband, they continued to work to organize and execute the 2009 CRN Family Conference in Atlanta. Lorna, in fact, organized and ran the entire childcare program at that conference. No one would have blamed them if they had decided to back off and care for their families. But instead their dedication to CRN and our community led them to push on – we will never be able to thank them for their sacrifices and the gift they made on our behalf.
We have made an effort to include as many individuals with cystinosis as possible on the Board – self-advocacy is one of the most important skills we can nurture in our children. Additionally there are many others who serve on special committees and hold fundraising events. All are incredible individuals who have committed their lives and precious spare time to meeting the needs of the community, despite their own personal challenges. They will undoubtedly serve as a great inspiration to all of our children with cystinosis. They will serve as excellent role models for all of our children.

We have grandmothers, singles mothers, entire families and parents of multiple children with cystinosis serving on the CRN Board. We have individuals with demanding careers who carve out time from their families to give of their talents. None are compensated for their time and many contribute significantly financially. All are dedicated to helping others with cystinosis find their way and improve their lives. We are blessed by their generosity and they inspire me every single day to keep pushing forward.

Outside of our circle of cystinosis families and caretakers, there are also many inspirational stories. Advocacy groups like the National Organization for Rare Diseases and Genetic Alliance continue to push the agenda of rare and genetic disease recognition and treatment forward – both organizations have been of great assistance to CRN during its formation and in helping us meet new challenges.

Marybeth Krummenacker and I recently attended the Genetic Diseases of Children conference in New York. There were hundreds of individuals there who run patient advocacy groups for hundreds of different diseases. Each session provided incredible stories of bravery, perseverance and optimism for us all to aspire to. Especially moving was the story told by NFL Hall of Fame Quarterback Jim Kelly. His son was diagnosed with Krabbe Leukodystrophy (KL), an inherited fatal nervous system disease. After his son passed away from the disease, he and his wife decided to use the resources at their disposal to start a foundation to seek out a treatments and a cure and support the KL community. Further, they saw the unmet need for increased and standardized newborn screening throughout this country, not only for his son’s disease, but for many others. He has worked tirelessly to increase newborn screening, work that has undoubtedly saved hundreds of thousands of children from the consequences of untreated, life-threatening diseases. His passion for his mission was incredible, an inspiration to all in attendance.

The concept of inspiration is one that we consciously integrated into the upcoming CRN Family Conference to be held this July in San Francisco. Cindy Abbott will be kicking off this year’s conference by sharing her story, “From Undiagnosed to Climbing Mt. Everest”. In 2007 she was diagnosed with the rare disease, Wegener’s Granulomatosis – three years later at the age of 51 and functionally blind in one eye she summitted Mt. Everest. She has gone on to become a strong rare disease advocate. Her bravery, tenacity and generous spirit have been an inspiration to me and I look forward to everyone meeting her at the conference.

Most of all, my son Jack inspires me every day. He is the happiest kid I know. He puts up with appointments, medicine, therapy and tutoring and does it with a smile on his face. He plays baseball and basketball and works hard to keep up with the other boys. He loves animals and has a gentle spirit. Jack insists on “capture and release” as the go-to method of bug catching in our house. Last week, Jack had braces put on his teeth. I couldn’t help but peek into the exam room now and again to check on his progress. You know, mother’s instinct at its best. He sat there patiently in the chair for over an hour with his mouth propped open, giving me the “thumbs up” sign each time he saw me. After the appointment, the technician shared that Jack was the “perfect patient”. I told her he ought to be, he has had plenty of practice. And in the days following he has hardly complained about the discomfort, reminding us that he deals with pain and discomfort each day of his life. He is more than an inspiration, he is my hero. I am certain Jack is representative of all of our brave children.

My friend gave me good advice in guiding me to write about something I felt passionate about. Ultimately, it is that passion that continues to inspire my work with CRN. I would bet that my fellow board members, advisors, committee members, donors and fundraisers would agree. The people I have met throughout this journey are precious to me, have changed my life forever and have become life long friends. In my last year as President of CRN, I hope to inspire our board members to accomplish as much as possible in advocating the interests of our cystinosis community forward. A personal thank you to each of you on an individual basis for your dedication and commitment to CRN. I hope you enjoy this edition of the Cystinosis Advocate and we look forward to seeing you in San Francisco!

Warmest Regards,

Christy Greeley

President and Executive Director
CRN Education and Awareness Update

By Paula Shal
Vice President-
Education and Awareness

CRN continues to look for new ways to educate the medical community and the general public about cystinosis. Each year representatives from CRN attend medical meetings and conferences. At conferences, we spread the word about cystinosis, attend workshops, and network. At medical meetings, we exhibit and network with medical professionals who are familiar with complications of cystinosis, such as kidney failure. This year we attended a conference we had never attended—The Genetic Diseases of Children—Advancing Research and Care meeting (see page 5).

Some of the benefits of educating others about cystinosis include:

- **Improved treatment for symptoms of cystinosis**
- **Earlier diagnosis**
- **Find new researchers**
- **Networking**
- **Family Support**
- **Fundraising**

The more we educate, the closer we will be to a cure for cystinosis. As part of the cystinosis community, there are ways everyone can educate the public about cystinosis including:

- **Educate yourself**—You will feel more comfortable talking to others if you are comfortable with information about cystinosis. The CRN website (www.cystinosis.org) is a great place to start. There is a library of articles about cystinosis, electronic copies of all our newsletters, and much more.
- **Print brochures**—The CRN website has an informative brochure geared toward the general public and a separate brochure for medical professionals. Take both brochures to doctor’s appointments. Put brochures for the public in the waiting room, and give the medical professional brochures to nurses, doctors, and anyone who will take one.

- **Contact your local media**—The media is often happy to include your story in the newspaper, online, or on TV. Sometimes all you have to do is ask. It may help to contact them around the time of an event—a kidney transplant, fundraiser, birthday, accomplishment, etc.
- **Social Media**—Utilize facebook, Twitter, etc. to share information about cystinosis as well as your own cystinosis story. Create a video about cystinosis and post it on YouTube. Be sure to use words in the title that will make it more searchable. Many people will not know to search for “cystinosis,” but you will reach a wider audience with words like “kidney” or “rare disease.”
- **Buy CRN Gear**—Cafepress.com sells CRN clothing and accessories. Wear this gear as a conversation starter for talking about cystinosis. Plus a percentage of your purchase goes to CRN!
- **Fundraise**—A fundraising event is a great way to educate about cystinosis as well as raise money for CRN. Make signs and print brochures for your fundraiser. Show your video or the CRN video about cystinosis. Talk to others about cystinosis. Enlist friends and family to do the same, so everyone at the fundraiser leaves educated about cystinosis. Many annual fundraisers do not bring in much money the first year, but attendees learn about cystinosis and spread the word. Subsequently, attendance and money raised increases the following year.
- **Attend the CRN Family Conference**—This is a great opportunity to educate yourself and to share your story with others. It is not just cystinosis families at the conferences. Volunteers, researchers, pharmaceutical companies, and others also attend.
- **Enlist Others**—Have your friends and family help you educate others by talking to others, distributing brochures, hosting fundraisers, visiting the CRN website, reposting cystinosis information on their social media pages, and much more.

It costs little to nothing to spread the word about cystinosis, and the dividends could be enormous. Cystinosis may always be a rare disease, but it does not have to be a “little known” rare disease. Spread the word!
CRN Attends Genetic Diseases of Children—Advancing Research and Care Meeting in New York, NY

Christy Greeley and Marybeth Krummenacker represented CRN at the "Genetic Diseases of Children: Advancing Research & Care" meeting in New York City, March 8 and 9, 2011.

The conference offered a comprehensive agenda presented through four plenary sessions and over 35 panel discussions covering five independent themes:

- Reaching An Early Diagnosis
- Sharing Family Experiences
- Optimizing Care And Coordination Of Services
- Children Living With A Genetic Disorder: Meeting Their Needs
- Accelerating Research

The Genetics Diseases of Children Conference was presented by the New York State Department of Health’s Wadsworth Center in conjunction with the National Institutes of Health's Office of Rare Diseases Research, the National Institutes of Health’s Office of Dietary Supplements, Genetic Alliance, the National Organization for Rare Disorders (NORD) and the Jeffrey Modell Foundation. For more information you can visit the meeting website at http://www.wadsworth.org/events/genetics/

As CRN representatives, we were able to network with many other rare, genetic disease non-profit groups, gathering information and advice on many of the above themes. There were several sessions dealing with transitioning issues, a topic CRN has focused on during the past year. Maya Doyle, CRN Professional Advisory Board member, was also able to attend and was able to gather much useful information with respect to the transitioning guide CRN is in the process of compiling.

CRN to Sponsor Cystinosis Symposium at the 2011 Latin American Pediatric Nephrology Association Meeting

The Cystinosis Research Network will sponsor a one day Cystinosis Symposium during the 2011 Latin American Pediatric Nephrology Association Meeting October 26, 2011 in Sao Paulo, Brazil. The meeting is chaired by Dr. Vera Koch from Brazil and Dr. Leticia Belmont from Mexico, both of whom are heavily involved in treating patients with cystinosis and cystinosis research. The meeting will likely attract 500-600 pediatric nephrologists from Latin America. The symposium will provide an entire session dedicated to educating these physicians about the diagnosis, treatment and research of cystinosis. CRN will sponsor Dr. Galina Nesterova, NIH Clinical Cystinosis Fellow, to be a featured speaker at the symposium. The Cystinosis Research Foundation is a co-sponsor of the event. Both groups will have the opportunity to present patient and family perspectives to the professionals in attendance. For more information you can visit the meeting website at http://www.alanepe2011.com.br/index-nova-ing.php.
CRN Family Support Update

By Cheri Friend
Vice President-Family Support

The Family Support Committee, along with the conference subcommittees, is busy at work preparing for the upcoming CRN Family Conference to be held in San Francisco, CA on July 14-16, 2011. The agenda is filled with presentations covering the various topics related to cystinosis – everything from the history of cystinosis to past favorites – the medical and patient panels. Registration packets were mailed the week of April 11th – if you have not received yours, please contact Cheri Friend at cfriend@cystinosis.org. Registration is also available online at the CRN website – www.cystinosis.org. Click on Events > Conferences > 2011.

The Family Support Committee also has oversight of the Adult Care Excellence Initiative work that is currently in progress. The members of this focus group are hard at work finalizing the transition guideline that will be presented at the upcoming family conference. We are excited to be publishing this guideline for our community and believe it will be very useful to the members of the cystinosis community as they begin the process of transitioning from pediatric to adult care.

Looking forward to seeing everyone in San Francisco in July!

CRN Adult Care Excellence Initiative

By Colleen Hammond

The final report from the Cystinosis Research Network Adult Care Excellence Initiative committee’s Living with Cystinosis Survey is now available on the CRN website at:

http://www.cystinosis.org/filemanager/file/Living_with_Cystinosis_FULL_FINAL.pdf

The group has been working very hard over the past several months to gather the information necessary to compile their ultimate goal of a cystinosis pediatric to adult care transition guide. Maya Doyle, a Social Worker at Montefiore hospital in NYC who works with Dr. Kaskel and is a member of CRN's Professional Advisory Committee, wrote and compiled the data for the survey with assistance from Dr. Elenberg and the transition committee.

The survey attracted 150 participants when it was distributed in November and December of 2010 and has revealed some really interesting data on this topic. Maya is an incredible asset to our community and we thank her for her work on this project!

We also thank all of you who participated -- the findings from this survey will not only inform the transition guide in process, but will also help direct CRN's efforts in regards to fulfilling the needs of the adult cystinosis community. This information will also be interesting to the broader rare disease community and we hope to publish this data more widely so that others can learn from what we have found within the cystinosis community.

We look forward to presenting this data, along with the final transition guide at the CRN Family Conference in July.
Report from the National Organization for Rare Disorders (NORD)

By Marybeth Krummenacker

Serving as CRN Board member and Chairperson of the Advocacy Committee for NORD keeps me on my toes. NORD has been extremely busy since November with frequent information and updates coming in from Washington DC. The decisions made in Washington have long reaching affects for all of those living with rare diseases, such as cystinosis. If it weren’t for the Orphan Disease Act of 1983, the cystinosis community WOULD NOT have cystagon as a treatment for cystinosis. Your help is needed to ensure legislation like this continues to be in the forefront in Washington. Please reach out to your elected officials in each and every state and ask them to sign on to the Rare Disease Caucus. Plans are being worked out now for this first-ever Caucus, and there will be more to report as we get further into the details. NORD’s website (rarediseases.org) has a list of names and states that have signed on to the Caucus. If your representative’s name is not there, please contact him/her and ask them to sign on to the Rare Disease Caucus.

As part of my NORD leadership role, I attend monthly conference calls and receive frequent updates from the Executive Director and the Public Policy Vice President. Since last November’s elections, many hours have been spent building relationships and making critical contacts in Washington to make sure that the voice of the rare disease community is heard. There were many changes in Washington this past November, so it takes time to foster those new relationships......but I am happy to report it is happening every day. NORD represents over 30 million people living with rare diseases, so we have an opportunity to have a large presence in Washington DC.

Rare Disease Day was held once again this year on February 28. More than 500 patient organizations and companies signed on as Rare Disease Day Partners.....CRN was one of them. There was a lot of “buzz” surrounding the day all over the world, including symposiums, rallies, media coverage, film screenings, and more. To know that CRN is a part of that world wide advocacy day is phenomenal!

I will be participating in the NORD Corporate Council Meeting on May 16 as well as a Board of Directors meeting on May 18th. The NORD annual Partners in Progress Gala is coming up on May 17th, and CRN will have representation for that wonderful evening as well. It will be an extraordinary event with almost 400 people in attendance. Plans are being made to hold a Rare Disease Conference in October in Washington D.C. that will be sponsored by NORD, the FDA and the NIH.

I am very proud to represent CRN and the cystinosis community on NORD’s Board of Directors. I bring more than 22 years of experience of living with my daughter, Laura’s cystinosis challenges as well as advocating for her. I understand that it is a unique opportunity and huge responsibility to be the “voice” at the table for all rare diseases.

CRN Accepting Nominations for New Board Members

Have you ever thought about becoming more involved in your organization? Have you ever thought about becoming a member of the board of CRN, helping to shape our group for the future? Now is the time to get involved. We are now accepting nominations for new board members. The Board of Directors will vote on new members in July. The board meets 3-4 times a year by telephone conference call. Once every 2 years we have an inperson full board meeting that takes place in conjunction with the Family Conference.

CRN is looking for committed people who are willing to get involved and work to keep CRN the outstanding organization that it has become. Being a volunteer helps others, lifts your spirits and is very rewarding. If you are interested, please contact Karen Gledhill (kgledhill@cystinosis.org) or Christy Greeley (cgreeley@cystinosis.org).

“Service to others is the rent you pay for your room here on Earth.” (Mohammad Ali)
Clinical trials of RP103 continue to progress. The current Phase 3 trial is in the final stages of enrollment and is expected to end sometime this summer, with at least 36 subjects completing the trial. The primary endpoint of the trial is the steady-state white blood cell ("WBC") cystine levels of patients taking RP103, a unique delayed release cysteamine, dosed every twelve hours, compared to immediate-release cysteamine, which requires strict dosing every six hours. We are doing all we can to quickly enroll the final patients and complete the trial. A high quality, reliable data set to support our New Drug Application (NDA) filing is of prime importance. The company hopes to submit our NDA to the FDA this year.
CRN Research Update

By Elva Smith-Vice President, Research

Progress continues to be made in CRN’s search for improved treatments and ultimately a cure for cystinosis. The CRN 2011 Call for Proposals was issued in mid-March and we hope to receive many grant requests for funding for new and innovative research. The formal request and the grant submission guidelines can be found on the CRN web site at www.cystinosis.org. CRN and the cystinosis community sincerely appreciate the support and dedication of our researchers. The following is a summary of CRN’s current research grants:

Rosaleen Anderson, PhD, Sunderland Pharmacy School, University of Sunderland, "Proteomic investigation of cystinotic cells and the effects of cysteamine treatment". Grant awarded: 23 November 2010 for a term of 2 years in the amount of GPB 80,926.00 or $130,366 based on the exchange rate for 15 November 2010.


Leticia Belmont, M.D., Unidad de Genetica de la Nutrician, Instituto Nacional de Pediatria, Mexico. “Determination of Intraleucocytic Cystine by High Performance Liquid Cromatography (HPLC) in Patients with Cystinosis”. Grant Awarded: 5 November 2008 for 1 year (Fall ’08 – Fall ’09). Total award: $31,972.00. Initial Payment: $15,986.00 1 December 2008 .Presentation, citing CRN funding, was given at the IPNA meeting in NYC, 31 August 2010. Progress report rec’d Sept. 2010; letter of inquiry sent to Dr. Belmont. Balance remaining on grant: $15,986.00

Henk J. Blom, M.D., M.M.C. Wamelink, and E. Levtchenko, VU University Medical Center, Amsterdam, The Netherlands: “Newborn Screening of Cystinosis”. Grant Awarded: 5 November 2008 for 18 months (Fall 2008 – Spring 2010), Total Award: 45,000 euros (approximately $67,500.00). Initial Payment: 8 December 2008 for $15,380.26. Balance remaining on grant: 33,750 euros (approximately $52,120.00).


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CRN Research Update (Cont.)

**Dr. Ewa Elenberg**, Texas Children's Hospital, Houston, TX  "Quality of Life in Cystinosis Patients", Grant awarded 9-25-09 for 1 year (10-1-09 to 9-30-10) Total award: $21,000.00. Initial payment: 9 Oct. 2009 for $5,213 Check was made out incorrectly and subsequently voided. Check was reissued. Six month progress report rec’d 12 November 2010; 2nd payment of $7875.00 issued 1/11/11. Balance remaining on grant: $7875.00

I know many of you are wondering about status of FDA’s approval of Sigma-Tau’s eye-drops for cystinosis patients. Here is the latest information that I have received:

**About CYSTARAN™’s Manufacturing Delay**

In September 2010, the U.S. Food and Drug Administration (FDA) issued a complete response letter to Sigma-Tau Pharmaceuticals, Inc. for CYSTARAN™ (cysteamine hydrochloride ophthalmic solution), a therapy for the treatment of corneal cystine crystals in patients with cystinosis. The final approval has been delayed to allow the third party manufacturer of CYSTARAN™ to meet additional standards of production requested by the FDA. While the FDA’s questions about manufacturing are impacting the timeline for product availability, Sigma-Tau Pharmaceuticals, Inc. is committed to the cystinosis community and is diligently working to reach the final stage of the product’s approval process. For more information regarding CYSTARAN™ and its pending FDA approval, please contact Lesli King at lesli.king@sigmatau.com.

CRN Fellowship Update

By Galina Nestrova M.D.

I was privileged to receive a CRN fellowship to be trained as an expert in nephropathic cystinosis at the NIH in order to serve the cystinosis community.

For the past year I continued clinical evaluation and treatment of cystinosis patients with a focus on early detection of complications in order to prevent long-term disabilities and to monitor appropriate treatments to preserve patients kidneys function. In regard to education of the medical community: I gave grand rounds to the university hospital doctors on the diagnosis and treatment of nephropathic cystinosis; also two medical publications were written: one for Netter textbook and, the second, pending an education article on cystinosis in the Pediatric nephrology journal. I continue to take an active role in teaching and educating the new genetics fellows at NIH on cystinosis. My major long-term research project has been initiated, with a focus on Fanconi syndrome and evaluation of cysteamine in alleviating Fanconi syndrome symptoms, as well as helping on developing newborn screening for cystinosis.

One of my most important goals is to improve the diagnosis of cystinosis in children by working with nephrologists and emergency medicine specialists who are often the first to see them.

I am very grateful to CRN, my current institution, my mentor Dr. William Gahl, to many families affected with cystinosis whom I offered and provided counseling on medical problems and concerns, for the opportunity to gain significant research exposure, clinical insight, and helping me to foster future collaborations throughout my career in the field of cystinosis.
Mitchell Smith Featured in Carefree Chronicle

Carefree Chronicle is a publication of Emory University/Children’s Healthcare of Atlanta. Mitchell Smith is enrolled in the delayed-release cysteamine study at Emory and was featured in the cover story of the November/December, 2010 issue. The article features Children’s pediatric research efforts and has been reprinted below with permission from Children’s Healthcare of Atlanta.


At only 7 years old, Mitchell Smith doesn’t understand his vital role in fulfilling the Children’s Vision for Research, but he definitely appreciates the entertainment value.

“At the clinic, Dr. Greenbaum juggles for us! The nurses play with us…they’re really nice,” he said. “And, I get to see other kids with cystinosis, like me.” What Mitchell won’t realize until he’s much older is that his clinical trial participation is advancing the leading edge of medical science—for his own rare kidney disorder, and for other children around the world who suffer from it. So, while he innocently delights in his doctor’s juggling act, Mitchell is also helping Children’s make history.

As part of Vision 2028, Children’s is developing a robust pediatric research enterprise to enable us to reach our goal to be one of the top pediatric research hospitals in the country. This year, we’ve opened seven centers dedicated to various areas of research. In 2011, we’ll open five more, two of which are focused on connecting bench research to bedside treatment—or, as in Mitchell’s case, out of bed and living the life of a healthy 7-year-old.

“2011 is the year all of our research centers will really come together. This past year we started seven outstanding new bench-to-bedside research centers attacking important diseases or research problems,” explained Paul Spearman, M.D., Chief Research Officer. “Our new Clinical/Translational Research Center and Center for Outcomes Research and Public Health will enhance research opportunities in all of our clinical service lines by supporting cutting-edge outcomes research and clinical trials science.”

The Children’s Clinical/Translational Research Center

The CCTRC will provide support to investigators from across the system performing drug trials and other interventions. Children’s already has a large group of expert clinical trials coordinators and research nurses, an accredited institutional review board, and other excellent resources for the performance of clinical trials. The CCTRC will organize these resources further, assist physician-investigators with the planning and performance of clinical trials, and provide services such as informatics, seminars, and statistical support. The Pediatric Research Trials Unit, located at Children’s at Egleston—and where Mitchell sees Dr. Greenbaum perform—is just one component of the CCTRC that supports cross-Site studies.

The study in which Mitchell participates involves testing a new drug that preserves his kidney function, prevents damage to other organs, and dramatically delays the need for a kidney transplant. And it can be administered much less frequently than his existing medication. “We were the first in the world to offer this study and are now one of just three centers in the U.S., so families travel here from many other states, as well as Canada,” said principal investigator Larry Greenbaum, M.D., Ph.D., who is also

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Mitchell Smith Featured in Carefree Chronicle (cont.)

At the Children’s Clinical/Translational Research Center, Mitchell and Mason take a break during their treatments with Dr. Greenbaum, who monitors them during their clinical trials and provides entertainment as well.

In pediatrics, many treatments aren’t as well studied as they are in adults. In many cases, if we weren’t conducting research, these treatments would never see the light of day.

Mitchell’s mom, Lorna Smith, would have taken him anywhere to receive the life-changing medication, but thanks to Children’s, the Lawrenceville resident didn’t have to. “I feel so blessed to have Children’s here and to be involved in the study. By helping Mitchell, this medication has tremendous potential to improve the quality of life for our whole family. Plus, everyone on the research team is an expert in their field, and they treat us like family,” she said. “It’s also quite gratifying to help pave the way for better lives for other kids like Mitchell.”

Center for Outcomes Research and Public Health

Like the CCTRC, CORPH will create research connections across clinical divisions. But at CORPH, the research itself takes a different form, focusing on things like improving quality of care delivery, studying environmental impacts on health, and determining how best to care for chronically ill children as they become adults.

“We’re very excited about how CORPH is taking shape,” said Ann Metten, Ph.D., Director, CORPH, and Professor of Pediatrics at Emory. “These two new centers create an important bridge from bench research to bedside knowledge means we gather insights and best practices that all children’s caregivers can apply to their patients. And through compassionate use guidelines, we’ve literally been able to make ‘miracle drugs’ available to non-trial patients,” said Dr. Greenbaum. “Our local patients often have early access to advanced therapies, simply by virtue of them being available through the CCTRC.”
Mitchell Smith Featured in *Carefree Chronicle* (cont.)

application, and will help put the work of all Children’s researchers on the national map."

One example of CORPH’s research involves the study of families and patients with cystic fibrosis. “With any highly complex, chronic disease, it’s important to recognize all the elements that either help us deliver quality care or stand in the way,” explained Michael Schechter, M.D., M.P.H., Director, Children’s Cystic Fibrosis Center. For example, there’s a lack of data about the prevalence and effect of normal health problems on the outcome of CF. “We know that when parents have difficulty coping with their child’s disease, there is a negative impact on the child’s outcome,” said Dr. Schechter, who is also Section Chief of Pulmonology, Children’s at Egleston. “Right now, we’re studying the prevalence of mental health issues in CF patients and their parents, which will lead us to causes and help reveal interventions to improve patient outcomes.”

Other recipients of CORPH seed grants are studying the use of health information technology (HIT) to help childhood cancer survivors transition from adolescence into adulthood, the use of web technology to integrate newborn screening data; and the impact of air quality on patients with asthma—one of the top reasons for admissions into Children’s.

"As we identify the most effective strategies, we not only improve care, but become more cost effective," said Dr. Mertens. “The results of this type of research are highly practical. In just a few years, it will be fascinating to see what revelations our researchers have uncovered.”

**Positioned for success**

As the country’s largest pediatric medical center, Children’s is well-positioned to become a leading research center. “With our three campuses combined, we offer researchers larger patient volumes than any other U.S. hospital,” said Kristine Rogers, Director, Clinical Research. “Investigators can study a tremendous volume of children here, so their studies can be larger and more effective. And our outcomes data is rich enough to yield meaningful results. Most importantly, high volumes mean researchers can find answers faster.”

Also attractive to researchers: Children’s is equipped to perform high quality clinical trials, with some of the best protection for patients. And we have top-notch electronic medical record technology to support data collection and analysis. Additionally, our relationships with other research organizations, like the Centers for Disease Control and the Georgia Institute of Technology biomedical engineering program, are a real draw.

"As we use our resources and relationships to energize the larger medical community, our status as a well-respected research leader is growing," said Dr. Spearman. “And as momentum increases in each area of research, we’ll attract the critical mass of people needed to successfully attack the problems we’re trying to solve. Of course, ultimately, the real beneficiaries are our patients.”

For Mitchell, research at Children’s has given him the ultimate gift: the chance to be a kid again. "It lets me play more!" he said.

And that’s what really matters. ☀️
All of us in the Cystinosis community are well aware of the medical definition of Cystinosis. Within a year or two of diagnosis, most of us can rattle off symptoms, statistics, lab results, medication doses as well as any doctor or nurse. But the human definition, the one we all carry in our hearts, the one that forever becomes the before and after in our lives is so much harder.

Our son, Steven turned 23 last February 13th. We celebrated with friends and family at his favorite restaurant in Ann Arbor. It marked the 22nd year of our journey with cystinosis. He was diagnosed in July, 1989 at eighteen months of age after a seven month search for answers for disturbing symptoms we all know by heart—severe failure to thrive, electrolyte imbalances, constant vomiting, poor appetite, kidney failure, anemia, unquenchable thirst, and massive urination. The image of that little boy is engraved in my soul as clear today as it was then. The plump, little sixteen pound four month old and the Stevie at eighteen months who weighed less then fifteen pounds, and looked like a preemie or concentration camp victim, huge blue eyes, translucent pale skin that hung in folds and the wisps of blond hair that fell out in clumps. In spite of it all, we were happy that day. We finally had the name of something to fight.

He started on phosphocysteamine in September, 1989. It would be six years before Cystagon was FDA approved. The early years were tough—many hospitalizations for dehydration, septicemia, encephalitis, malignant hypertension, electrolyte imbalances, and anemia. There were frequent labs and doctors’ visits. He had a G-tube placed in January, 1990 which was a Godsend for medications and nutritional supplements. It was not removed until he was nine years old. Very slowly he improved and stabilized. He learned to swallow pills at age six at a cystinosis conference, an event that had me sobbing. I was well aware how momentous that ability would prove to be to a child who needed to swallow dozens of pills a day. There were ‘normal’ years too. Years filled with school projects, soccer, Cub Scouts, birthday parties, and family vacations. Steven taught us all the beauty and value of ordinary moments.

In 1999, at age eleven, he had a successful kidney transplant. That clearly marked the second stage of this long journey. His dad often jokes, “same disease, different meds.” For Steve, post transplant has truly been an improvement in his quality of life. He grew ten inches within two years of transplant. He did have a few minor rejection episodes early on and one gastritis episode, but overall has been very stable. He found a love of drama in high school and performed in many plays over the years including two since he graduated in 2007. He also loves music, plays guitar and frequently performs with friends at venues nearby.

He is currently a college student at Eastern Michigan University majoring in Graphic Design. He continues to take about 30 pills a day and uses eye drops several times a day to dissolve the corneal crystals. For the past three years he has been managing his own health care needs as he successfully transitioned from pediatric to adult doctors. He is doing a fine job. Steve is an inspiration to us and many who know him. He pursues his dreams and goals with a spirit and determination that cystinosis cannot undermine.

Our family has found many gifts during this journey. One of the most precious to us has been attending many family conferences over the years and meeting other families who walk a similar path with Cystinosis. Friends we’ve made over the years have truly become family to us. We look forward to renewing those relationships in San Francisco in July.
Kathleen was born in October, 2000—a thriving 9lb 2oz baby. She was enormous (!), beautiful, and as far as we knew, healthy. Although she was colicky, she thrived her first 6-7 months of life. She breastfed well and met all of her developmental milestones. We had no worries. But at about 8 months of age, things changed. Our first clue that something was wrong was her voracious thirst and incomprehensible urine output; she was also slow to pull up and became very constipated. Then, at about 10 months of age, she completely lost her appetite. Her weight dropped, and her thirst became so extreme she was frantically drinking the bath water only to vomit it right back up. She was admitted to hospital at 11 months of age and found to be spilling protein and phosphate in her urine; her serum phosphate levels were very low. Being in the medical field I immediately researched all the possible diagnoses…I will never forget reading about cystinosis that evening in the hospital library. I knew what she had immediately.

It was not long after that we received confirmation of her diagnosis. Like so many families, we were devastated but also relieved to have a diagnosis. Our saving grace was receiving an email and video from Colleen Hammond and CRN. Kathleen was still in hospital and not doing well, but Colleen, someone who had firsthand experience with this illness, gave us so much hope for our baby girl.

And hope we did. We hoped through investigations, the g-tube surgery, the daily q6h medication administration, the daily vomiting and readministration of meds, the multiple admissions to hospital for dehydration, the initiation of growth hormone and cysteamine eye drops, and now we hope that soon Kathleen will start swallowing pills and be eligible for the RP103 delayed release cysteamine study. And as we hoped, our local community, friends and family rallied around us, supported us, and hoped with us.

In 2002 we hoped after the birth of our second daughter, Sarah and were relieved to find out she did not have cystinosis. We are so fortunate now to have two great little girls who are wonderful companions to each other.

With time, our hope has grown into confidence that Kathleen will enjoy a “normal” life. This confidence comes from seeing Kathleen grow into a wonderful, happy and compassionate person. It comes from seeing her enjoy normal childhood activities; skiing each winter, swimming, successfully surfing this spring (she was the first in our family to get up on the board!), having sleepovers and play dates, singing in a choir, dancing, and playing piano. She has not had a hospital admission in over 2yrs, and although she is still prone to vomiting, it is much less frequent than it used to be. Our confidence also comes from witnessing other children and adults with cystinosis thrive and succeed in life and from knowing how much effort and research is going into helping children and adults with cystinosis—whether it is finding a cure or providing better treatment options for them. We are so grateful to CRN, CRF and now CARE in Canada for the amazing amount of work they have done in supporting families affected by cystinosis. We feel incredibly fortunate that so many people are working hard to make our little girl’s life easier and better.

Every day we wish for a cure. Every day we wish for something to make Kathleen’s life easier, but every day we are thankful for what we have and for the support we receive from our local community and our cystinosis community.
CRN Development Update

By Jen Wyman-Vice President, Development

"Although the world is full of suffering it is also full of the overcoming of it."
Helen Keller

As I write this article I am also watching the news on the aftermath of the earthquake and tsunami in Japan. It is astounding to me how the Japanese people are keeping their dignity and grace in the face of adversity. It is a reminder of how to live our lives... whether facing a new day amongst devastation and loss or a new day facing ongoing struggles with cystinosis. One thing remains true..."the human spirit is stronger than anything that can happen to it" (George C. Scott).

The Cystinosis Research Network has done some pretty incredible things in its 15 years. I have been affiliated with it since 2006, when my daughter was diagnosed. Twice a year I write an update for the newsletter and each time I am amazed by the ongoing and promising things that are taking place, both currently, in the near future, on the horizon and even OVER the horizon. It’s what keeps our hope alive.

In March, The Krummenaker family held their Millridge Dinner Dance with over 350 people in attendance, and the cystinosis community was well represented. They raised over $20,000 for CRN. There is a tremendous amount of effort that goes into an event of this magnitude, but the result is more than a dollar amount; It’s a heartwarming, inspiring night to remember.

May and June will bring us more in the way of successful fundraising with big events: The abeXchallenge, created by the Julian family, honoring their daughter Kenadee. The Wyman’s 5th Annual Cystinosis Fun Run, which honors our daughter, Kacy. The C.H. Robinson Golf Outing in Utah, honoring Tahnie Woodward, which will celebrate its 5th year, and Jack Greeley’s birthday letter. These four events raised over $125,000 cumulatively in 2010.

Later months will bring us more letter campaigns, dance-a-thons, sporting events, and Facebook fundraising. All of which bring us closer to our goals of making the lives of those living with cystinosis richer and more promising.

The Cystinosis Advocate is full of stories and articles about fundraising events, family support, medical research studies and updates, and conference information. It is a culmination and a representation of all the hard work that CRN does to support those that live with cystinosis. So many of you who are close to my family remark of these beautiful and comprehensive newsletters we mail out. You receive it because at some point you were touched by someone, within the very small cystinosis community, and made a contribution. Whether you were touched directly or indirectly makes no difference. It does not go unrecognized and providing you this material shows you what your donation dollars do for CRN.

Plain and simple, the Cystinosis Research Network is a not for profit organization. It depends heavily on fundraising and donations to exist and to educate and support the families. Thank you!
## 2011 Fundraising Calendar

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<th>Date</th>
<th>Event</th>
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<td>February 7</td>
<td>2nd Annual Eastland High School Cystinosis Awareness Week</td>
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<td>Sarah Larimore</td>
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<td>November</td>
<td>Jake Krahe Letter Campaign</td>
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“The best work is done with the heart breaking, or overflowing.” Mignon McLaughlin
On Saturday March 5, 2011 the 6th “Miracles at Milleridge Dinner Dance” was held at the Milleridge Cottage in Jericho, New York. The event was an overwhelming success with over 330 people in attendance. The night began with a silent auction that included sports memorabilia such as autographed footballs and baseballs to a huge selection of original and autographed photos. Over 58 baskets were also raffled off with items ranging from bottles of wine to trips to Wyoming and North Carolina. The festivities also included a 50/50 raffle.

The evening began by Marybeth Krummenacker introducing and thanking her committee of family and friends and acknowledging the fact that the evening is only successful because of all of the help and support family, friends, and everyone in attendance. There were 7 families representing 6 different states in attendance. A special presentation was made to Dr. Frederick Kaskel by Marybeth and Laura Krummenacker. Dr. Kaskel has been Laura’s pediatric nephrologist for more than 22 years, and he was presented with a framed collage of photos of Laura throughout the years. Laura is taking that very BIG step of finally transitioning into the adult world of nephrology this year so it was time to say a special thank you to Dr. Kaskel for being at Laura’s side as not only a doctor but also a friend. It was a very special, emotional moment for all.

Another special moment was when Marybeth asked Tahnie Woodward to bring her 13 month old daughter up for a very special introduction....Miss Sookie Sierra Boyer was introduced as one of the very special “miracles” of the cystinosis community. Sookie took it in stride and was certainly up for a party! The evening was a huge success with over $20,000 being raised for the Cystinosis Research Network!

Miracles at Milleridge Raises $20,000+ for CRN!

Laura Krummenacker, Tahnie Woodward, Sookie Boyer, and Marybeth Krummenacker at Miracles at Milleridge Dinner Dance

Pam Woodward and granddaughter, Sookie

Moms with Cystinosis, Tahnie Woodward and Emily Mello, and their kids, Sookie and Elijah

Christy Greeley, John Maccarone, Cheri Friend, and Colleen Hammond
Greeley Birthday Letters Have Raised $150,000+ Over Nine Years

For the ninth year, the Greeley family has sent their annual appeal to friends, family and colleagues to raise money for CRN in honor of their son Jack's birthday on St. Patrick's Day. An excerpt from the letter follows. The full text of the letter can be found on the CRN website at http://www.cystinosis.org/filemanager/file/JackCRNletter2011.pdf.

"Last baseball season, Jack played on the Cubs in the local rec league. There was plenty of "thrill" and a fair dose of "agony" along the way. Dave coached third base for Jack's team. During one routine game, Jack got on base and managed to get over to 2nd base, which kids loved because rules allowed stealing of third base. Beyond the recessive gene for cystinosis that he inherited from both his parents, Jack also got lead feet from his Dad. It did not matter;; Jack was on second and he wanted to steal. While at his 3rd base post, Dave heard and saw a non-coaching parent lean over the fence and say: "Get the ball down to 3rd base...he is a slow runner." The first reaction – seriously – was for Dave to punch the guy in the face. Okay, not the best example for kids, so Dave gave the guy a death stare and (to paraphrase) said: "Hey, maybe the kid has a metabolic-genetic condition that does not allow him to run fast. Besides, since when is it appropriate to chime in about a kid's skill set?" He embarrassingly got the point. That sort of heartache is not uncommon for Jack, particularly coming from those who do not know him. He did not hear the comment, but his Dad felt it.

En route to finishing the regular season with a not-so-stellar 3-7 record, the Cubs were stuck in a dog fight with another sub-par team. Heading into the top of the final inning, the home Cubs held a 3-1 lead when they quickly allowed their opponent to score two runs to tie the game. As Dave headed out to coach 3rd base, he knew Jack was second to bat in the bottom of the sixth. He said a prayer: "Lord, PLEASE be there for the kid...help him to build some confidence...give him a moment." Jack's teammate batting first hit a routine ground ball, which in this league could easily result in a runner on base. A fielder scooped it up nicely, but overthrew first base...lead-off batter went to second and up came Jack. The pitcher threw a wild one, moving the runner to 3rd base. No outs, winning run on third, and Jack is at the plate! The drama built as the manager huddled up the entire infield telling them that they had to throw to the catcher to get the runner out if Jack hit the ball into play. Jack looked down at his Dad and basically said/thought: "What the heck is going on out there? I have never seen this before." There was actually tension. A few more pitches were thrown and a foul ball, leading to a full count. Jack, who is actually a pretty decent contact hitter, connected with the next pitch, hitting it solidly toward right field. Mind you, it was not Ruthian or a scene out of "The Natural," but it was a solid shot...now would it stay fair or would the outfielder catch it? Dave knew his duty...the runner needed to tag up...no matter what!!! Guess what? The ball was hit well and went past the right fielder's out stretched arm. While the kid on 3rd advanced home, Jack got to first base, turned around, and with the biggest smile any 10 year old had ever showed, yelled, "YES!," raising his arms in Rocky Balboa triumph. Cubs win! The 3rd base dugout cleared and the team ran to Jack in celebration. A game winner for Jack!

Last year's letter raised nearly $30,000 -- in the past nine years the letters have raised over $150,000! The Greeleys are thankful for the blessing of so many generous and supportive friends and family and a community that has supported them so enthusiastically over the years.
Jeans for a Cause Raises $1200+

Carl Schleuder’s employer, Ricardo, Inc., chooses a charity each month to be the recipient of funds raised from “Jeans for a Cause.” Jeans for a Cause allows employees to wear jeans on Fridays if they donate $1.00 to the selected charity. The company matches all contributions.

Cystinosis Research Network was the charity chosen in March. Ricardo, Inc. employees raised $620 in honor of Steven Schleuder, Carl’s son. The company matched the $620 for a total of a $1,240 donation!

CRN would like to thank Carl and all Ricardo, Inc. employees who contributed.

CRN is also saddened to learn Carl Schleuder’s mother, Mary A. (McGuire) Schleuder passed away on March 21, 2011. Her memorial is Cystinosis Research Network. Approximately $200 had been donated at the time the newsletter went to print.

5th Annual Shoot for the Cure Raises $1100

After selling a few pancakes and raising $1100 for a worthy cause, the Whitesboro Bearcats from Whitesboro, TX stepped onto the basketball court for the 5th annual Shoot for the Cure Night in honor of Mason Reed. Mason is the nephew of the head boy’s basketball coach, Shad Reed. Mason is in third grade and is participating in the Delayed Release Cysteamine study at Emory University in Atlanta. Shoot for the Cure has raised over $6000 in the past 5 years. The Reeds would like to thank everyone who helped make this event a success. There were a lot of people behind the scenes who sacrificed their time. There were numerous businesses who donated to the cause as well. The Reeds are proud to be a part of a great community that cares for kids.
Tahnie Woodward Uses Facebook and Twitter to Raise Money for CRN in Honor of her Daughter’s 1st Birthday

By Tahnie Woodward

Online fundraising can seem daunting at first, but a few tips and a lot of persistence can go a long way. In the past, I have used the 'Causes' feature through Facebook for fundraising but the most recent time I really committed to raising a hefty chunk of money for CRN. I started a birthday wish in honor of my daughter, Sookie, turning 1. I added some information about Cystinosis to the page, for anyone who wasn’t familiar with it. I have to say, I truly believe the reason this time around was much more successful than previous attempts is that I was determined and posted many reminders. I made a goal of $1,000, even though secretly I would have been over the moon with $500. A good tip is to make your goal for higher than you think you should. I think this gives great initiative and inspires you to work harder. In addition, I made sure to highlight the notion that even $10 would make a huge difference. I think sometimes people have the wrong idea that the donation needs to be a substantial number. I reached out to many strangers this time around as well; I posted about it on my Twitter account and Facebook. I also emailed a few people with popular blogs that I enjoy reading. People are much more willing to help than we often think! I made sure to post frequent updates on Facebook with the growing total as well as thank yous to those who had already donated. Something else I didn’t think of before, a few of my relatives preferred to donate offline through checks when I asked them about it. I told them that was no problem! The combination of offline and online donations reached a grand total of $1310 for a fundraising venture that didn’t require a momentous effort and did not have any expenses involved. Overall, I think this is a wonderful avenue for fundraising!
Paula Shal Hosts Facebook Fundraiser for CRN to Celebrate her 40th Birthday

Paula Shal hosted a fundraiser using facebook Causes for her 40th birthday on Dec. 8, 2010. She was surprised at how easy and effective it was. Facebook walks you through the process, lets you write a birthday fundraising wish, sends out reminders, and allows you to post personalized “thank you” notes to your donors. Facebook Causes also allows others to post your fundraising wish. Paula received some donations from “friends of friends” she didn’t even know! In addition to the facebook campaign, Paula wrote in her Christmas cards asking people to donate. In all, Paula raised over $1600 for CRN with no investment and very little effort! Paula would like to once again thank her friends, family, and wonderful husband who not only supported her fundraiser but have supported her through the ups and downs of cystinosis and helped her grow as a person.

Below is what was written as her facebook fundraising wish:

“When I was a teenager I didn't think I was going to live to be 25. Now I am 40. There is no big party, no big gift, and no big trip planned. After I received my 4th kidney transplant last fall, Jim and I decided since we were celebrating our 10th anniversary and my 40th birthday, that we would just treat every day of 2010 like a celebration. We vowed to try to take advantage of "once in a lifetime" experiences and to appreciate each day we have been given.

I am so grateful for the miracles and blessings in my life. I have benefited from modern medicine, brilliant doctors, generous loved ones, and a little bit of luck. I want to show my appreciation by making a donation to the Cystinosis Research Network, and I am hoping you will help me. Cystinosis is the disease I have, and while there may not be a cure in my lifetime, I want to make sure that there is a cure during the lifetime of the amazing kids I have met who fight cystinosis every day.

Cystinosis is a devastating cellular disorder that impairs every cell in the body. It leads to kidney failure, hypothyroidism, growth failure, requirement for feeding tubes, muscle weakness (including digestive and respiratory), severe sensitivity to light, diabetes mellitus, and neurological issues.

My birthday wish is in honor of my kidney donors--Mom, brother Brian, husband Jim, and friend Sue. If you are able, please consider a small donation to the Cystinosis Research Network in the month of December. To see the great work CRN does, please visit [www.cystinosis.org].

Do you want to connect with cystinosis families in YOUR area?

Visit http://www.cystinosis.org/families-in-your-area. Send your contact info to the e-mail listed to learn who is in your area. It’s that easy!
Eastland High School in Lanark, IL hosted the 2nd Cystinosis Awareness Week in honor of Paula Shal during the week of February 7. The week included hat day, in which the students paid $1.00 to wear a hat to school; a raffle, which included an American Girl Doll and Shake n’ Go Extreme Extreme Race Track; taco supper before the boy’s basketball game; and a “Pie-in-the-Face” contest, which targeted senior girls and boys basketball players.

Paula was scheduled to go speak to the student body at Eastland as she did last year, but extreme blizzard conditions in the Midwest forced her to cancel the 2.5 hour trip.

The event was sponsored by the EHS Student Council led by Marybeth Landherr. Paula Shal’s niece, Logan Feltmeyer, is a senior at Eastland and a member of the student council. Marybeth is a friend of Paula’s family who has done a great job at raising awareness for cystinosis over the past two years. The fundraiser raised just under $900.

"It was fun to see the whole community come together again to raise money to try and find a cure for this debilitating disease," was Logan’s response to the amount of money raised.
Dear Family and Friends,

It will be nearly 3 years since we hosted A Festival of Hope! Time has flown by and so many of you have asked when are we going to do it again. Well mark your calendars for Thursday, September 15, 2011!

Our family is thrilled to announce that the A Festival of Hope benefit will take place once again at Carlucci restaurant. All proceeds will go to support the Cystinosis Research Network (CRN). CRN is a private, non-profit 501c3 corporation, Federal Tax ID 04-3323789. Go to [www.cystinosis.org](http://www.cystinosis.org) for more information.

**What:** A Festival of Hope in honor of our children Alexis & Evan LeBeau who both suffer from cystinosis.

**Where:** Carlucci Restaurant 1801 Butterfield Rd, Downers Grove, IL 60515 ([www.carluccirestaurant.com](http://www.carluccirestaurant.com))

**When:** Thursday, September 15, 2011

**Time:** 5:30-9:30pm

**Cost:** $60 for 1 ticket or $100 for 2 tickets

**Contact:** Dana Drury-Brabeck, Event Director, 630-452-9884, [afestivalofhope@gmail.com](mailto:afestivalofhope@gmail.com)

**Website:** [www.afestivalofhope.com](http://www.afestivalofhope.com).

All guests will enjoy unlimited food and 3 hours of open bar, as well as live entertainment in the outdoor courtyard. The silent/live auction items and raffle will also not disappoint which included a flat screen TV, ipods, VIP concert tickets, custom made furniture, Bears/Blackhawks tickets, autographed sports memorabilia and much more!

For more information or to donate items for the auction, please contact Event Director, Dana Drury-Brabeck. Thank you very much for your time and efforts. We are appreciative and grateful for all of your contributions and look forward to your continued support. See you in September!

Blessings to All,

Brett and Brittney LeBeau
All Golfers Welcome to C.H. Robinson Cystinosis Charity Golf Tournament

Presents the 5th Annual

Cystinosis Charity Golf Tournament

The 5th annual Cystinosis Charity Golf Tournament will be June 11th at Thanksgiving point—Lehi, Utah. Shot gun start will tee up at 9:00 AM with registration, the putting contest and driving range opening at 8:05 AM. This a top fund raising event for Cystinosis Research Network.

Sponsorship Opportunities:

Event Sponsor:  Only One Left!  $2,000

- Name / Logo recognition
- 2 – Foursomes for the event
- 8 – Personalized gift for your players
- 1 – Professional banner with your company logo at the event!
- 2 – Foursome golf certificates at Thanksgiving Point Golf Course
- 8 – Putting challenge tickets

Reserve one of the following “Events”: Early Bird Breakfast, Lunch, Driving Range, or Drink cart

Hole Sponsor:  Only Two Left!  $1,000

- 1 – Foursome for the event
- 4 – Personalized gift for your players
- 1 – Large professional banner with your company logo at the event!
- 1 – Foursome gift certificate at Thanksgiving Point Golf Course
- 4 – Putting challenge tickets

Reserve one of the following “Hole events”: Longest Drive Man, Longest Drive Lady, Pay the Pro, Better Your Ball, Longest Putt, Final Resting Place, or Hole-in-One Give-away

Golfers:

Foursome Includes:  $500

- 1 Foursome, 4 event bags, early bird breakfast and lunch, 4 putting challenge tickets

Twosome Includes:  $300

- 2 Golfers, 2 event bags, early bird breakfast and lunch, 2 putting challenge tickets

Singles Includes:  $200

- 1 Golfer, event bag, early bird breakfast and lunch, 1 putting challenge ticket

Donations:

Donations are always welcome for the raffle. We are also allowing Sponsors to supplement some of their cash contribution in exchange for prizes to be raffled off at the event. Please contact Tim Miller at 801–768–2232 X 9 for information.
The Cystinosis Research Network has established a scholarship fund to provide supplemental financial assistance to a student diagnosed with Cystinosis who is enrolling in a regionally accredited collegiate or vocational program, or who is currently attending a post-secondary school. The scholarship award, $1000, is awarded contingent upon the winner's acceptance to an accredited college, university, or vocational program, or documentation of continued enrollment, and will be payable to the educational institution to be applied toward tuition. An application form is available on the CRN website at www.cystinosis.org/scholarships. For more information or to have an application mailed to you, please contact CRN at 1-866-276-3669 or info@cystinosis.org.

APPLICATION PROCEDURE:
Each applicant must submit:
1. Application form
2. Documentation/verification of Cystinosis (e.g. Letter from physician)
3. An official copy of high school transcript
4. Two letters of recommendation from current teachers/faculty members and/or counselors regarding applicant's scholastic aptitude and personal qualifications.
5. An essay of 500 words discussing the applicant’s personal and educational goals.

JUDGING CRITERIA: The essay will be judged on the basis of rationale, grammar, and comprehension. Transcripts and letters of recommendation will be considered in the final decision with Grade Point Average (GPA), courses taken, and class standing used as part of the evaluation. The Cystinosis Research Network Board will establish an independent judging panel to evaluate and rate the applicants. The decisions of the judges are final.

Finalists may be interviewed before selections are made.

DEADLINE FOR APPLICATION: Application and all accompanying documents must be received at the Cystinosis Research Network office postmarked by August 15, 2011. FAXES OR E-MAILS WILL NOT BE ACCEPTED.

PREPARING APPLICATION PACKAGE: Each application packet must include a complete application (original or photocopied—go to www.cystinosis.org to download), required documentation materials and essay, on 8 1/2” X11” white paper. Send all materials in a single, flat package. All application documents become the property of the evaluation committee.

MAIL APPLICATION PACKETS TO:
CRN Scholarship Committee
c/o Paula Shal
1718 Briarwood Dr.
Johnsburg, IL 60051
2011 CRN Sierra Woodward Sibling Scholarship

The Cystinosis Research Network has established a scholarship fund to provide supplemental financial assistance to a student who has a sibling diagnosed with Cystinosis who is enrolling in a regionally accredited collegiate or vocational program, or who is currently attending a post-secondary school. The scholarship award, $1000, is awarded contingent upon the winner’s acceptance to an accredited college, university, or vocational program, or documentation of continued enrollment, and will be payable to the educational institution to be applied toward tuition.

An application form is available on the CRN website at [www.cystinosis.org/scholarships](http://www.cystinosis.org/scholarships). For more information or to have an application mailed to you, please contact CRN at 1-866-276-3669 or info@cystinosis.org.

APPLICATION PROCEDURE:

Each applicant must submit:

1. Application form
2. An official copy of high school transcript
3. Two letters of recommendation from current teachers/faculty members and/or counselors regarding applicant’s scholastic aptitude and personal qualifications
4. An essay of 300 to 500 words discussing the applicant’s personal and educational goals and also the pro and/or cons of how Cystinosis has affected your life

JUDGING CRITERIA: The essay will be judged on the basis of rationale, grammar, and comprehension. Transcripts and letters of recommendation will be considered in the final decision with Grade Point Average (GPA), courses taken, and class standing used as part of the evaluation. The Cystinosis Research Network Board will establish an independent judging panel to evaluate and rate the applicants. The decisions of the judges are final.

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MAIL APPLICATION PACKETS TO:

CRN Sibling Scholarship Committee

c/o Marybeth Krummenacker

54 Smith St

Hicksville, NY 11801

Kim, Bree, and Jake Forrester
Join the Cystinosis Research Network

Get connected! Stay informed! Together we can find a cure!

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 Cystinosis Research Network (CRN) is a volunteer, non-profit organization dedicated to advocating and providing financial support for research, providing family assistance and educating the public and medical communities about cystinosis. 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:

“Dedicated to a Cure. Committed to our Community”...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 biannual CRN Newsletter, our very informative web page www.cystinosis.org, the 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.
- 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.

Cheri Friend
VP Family Support, Cystinosis Research Network
Join the Cystinosis Research Network Today!

<table>
<thead>
<tr>
<th>Immediate Family</th>
<th>$20.00</th>
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<tr>
<td>Extended Family / Friend</td>
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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:
Cystinosis Research Network
302 Whytegate Ct.
Lake Forest, IL 60045

*****************************************************************************
Name_______________________________________________________________
Street________________________________________________________________
City & State_______________________________________ Zip Code__________ Country ____
Phone__________________________Fax_______________________Email________________

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 two 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.

To join a support group, visit www.cystinosis.org.
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: 302 Whytegate Ave., Lake Forest, IL 60045

Name________________________________________
Street_______________________________________
City & State___________________________________ Zip Code_________________
Phone_________________ Fax___________________ Email_____________________

In Honor Of_____________________________________________________________
In Memory Of___________________________________________________________

You may send notification of my gift to: ______________________________________

Please check all that apply:
_____Friend
_____Individual with Cystinosis
_____Parent of Child with Cystinosis
_____Professional
_____Family
_____I am interested in volunteering for CRN. Please contact me.

Search the Internet Using GoodSearch and Raise Money for CRN

Every time you use GoodSearch.com to search the Internet, a donation is made to the Cystinosis Research Network! GoodSearch.com is powered by Yahoo!, so you get the same great results you get from most search engines. What is unique is that GoodSearch.com has developed a way to direct money to your selected charity with every click!

To get started, go to www.goodsearch.com. Be sure to select "Cystinosis Research Network" where it says "Who do you GoodSearch for?" Then search like you normally would! It costs you nothing.

CRN hopes you will give it a try and support Cystinosis Research. The more people who use this site for CRN, the more money is earned. So please tell your friends and family!

Make Online Purchases using GoodShop and Raise Money for CRN

GoodShop.com allows you to purchase through most online retailers, and a percentage of your purchase goes to the Cystinosis Research Network! There is no additional cost to you!

To get started, simply go to www.goodshop.com, choose CRN as your charity, and be sure to click through the link on the GoodShop page to get to your favorite retailer. It’s that easy!

GoodShop will donate up to 30% of your purchase to CRN. Some of the hundreds of retailers include: Best Buy, iTunes, Home Depot, Amazon, Barnes & Noble, Dell, Banana Republic, Macy’s, Target, Wal-Mart, Ann Taylor Loft, Chicos, Coldwater Creek, Christopher and Banks, American Eagle Outfitters, and many many more!
Identify the Cystinosis Research Network, Inc. as the agency you want to receive your contribution through the United Way Donor Choice Program.

<table>
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<th>Agency Name</th>
<th>The Cystinosis Research Network, Inc.</th>
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<tbody>
<tr>
<td>Non-Profit Tax ID #</td>
<td>04-3323789</td>
</tr>
<tr>
<td>Address</td>
<td>302 Whytegate Ct., Lake Forest, IL 60045</td>
</tr>
<tr>
<td>Telephone</td>
<td>1-866-276-3669 (toll free), 1-847-735-0471</td>
</tr>
<tr>
<td>Fax</td>
<td>847-235-2773</td>
</tr>
<tr>
<td>E-mail Address</td>
<td><a href="mailto:CRN@cystinosis.org">CRN@cystinosis.org</a></td>
</tr>
<tr>
<td>Web Page</td>
<td><a href="http://www.cystinosis.org">www.cystinosis.org</a></td>
</tr>
</tbody>
</table>

The local United Way organization will contact The Cystinosis Research Network via phone, fax, or e-mail to request we prepare and submit documentation verifying our status as a non-profit organization.

The Cystinosis Research Network prepares all necessary documentation and submits it to the respective local United Way organization.

The local United Way organization processes the documentation and sends a check for the aggregate sum designated for the Cystinosis Research Network.

The Cystinosis Research Network sends thank you/acknowledgement letters to recognize contributing individuals.

**Donate to CRN by Selling on eBay**

CRN is registered with MissionFish, the exclusive charity provider for eBay Giving Works. eBay sellers can now list items through eBay Giving Works and designate a percentage of the sales to go to CRN. The seller picks the percentage, and all money donated is tax deductible. eBay will even refund a percentage of listing and final value fees that is equal to the percentage sellers donate! Items listed with eBay Giving Works are given a special icon, so they stand out. Some sellers report 20-40% higher sale prices for the exact same item using eBay Giving Works. Give it a try, and be sure to tell established eBay sellers about this great opportunity to give to CRN!
Cystinosis is a rare disease that primarily affects children. Cystinosis is a genetic metabolic disease that causes an amino acid, cystine, to accumulate in various organs of the body. Cystine crystals accumulate in the kidneys, eyes, liver, muscles, pancreas, brain and white blood cells. Without specific treatment, children with cystinosis develop end stage kidney failure at approximately age nine.

**CRN Vision and Mission**

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

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