CRN Family Conference Set for July 14-16, 2011 in San Francisco, California

The Cystinosis Research Network is pleased to announce its 2011 Family Conference will be held July 14-16 at the Hotel Nikko in San Francisco, California. This will be CRN’s fifth family conference, with previous events held in Orlando, Florida (2003), Salt Lake City, Utah (2005), San Antonio, Texas (2007) and Atlanta, Georgia (2009).

Please plan to join us as we demonstrate how CRN is building a "Bridge to the Future" through our support of the Cystinosis community through Research, Family Support and Education and Awareness efforts. You’ll learn about new research findings, receive updates on cystinosis and rare disease organizations from around the world, meet and renew friendships with our inspirational families, have the opportunity to participate in research studies, and interact one on one with many of the world expert clinicians treating and researching cystinosis today.

Cystinosis Research Network Awards NIH Cystinosis Fellowship

The Cystinosis Research Network is pleased to announce that it has awarded its National Institutes of Health Cystinosis Fellowship to Galina Nesterova, MD. William A. Gahl, MD, PhD, of the National Human Genome Research Institute, will direct the three-year fellowship whose goal is to train an expert in nephropathic cystinosis at the NIH. The intention of the fellowship is to provide the next generation with an authority in the disease, a vitally important unmet need in the community. Dr. Nesterova, who received her initial medical training in Russia, currently serves as a Medical Genetics Fellow at the NIH, National Human Genome Research Institute. She has experience in clinical and bench research in general and biochemical genetics (diagnosis and treatment of inborn errors of metabolism) in particular. Her major interests are renal genetic disorders with focus on nephropathic cystinosis, Fanconi’s syndrome and nephrocalcinosis. Under Dr. Gahl’s mentorship, Dr. Nesterova hopes to accomplish a wide range of objectives during the fellowship including continuing clinical evaluation of cystinosis patients at the

Continued on page 3
Message from the President  
Christy Greeley

By Christy Greeley
“Bridge to the Future”

As we approach the holiday season, it is difficult not to reflect on the past year and give thanks for the many blessings we’ve received...new families diagnosed earlier, children receiving better treatment, adults living long and productive lives...much promising and innovative research occurring around the world...patients being diagnosed and treated appropriately in countries across the globe...new clinical specialists committing to a career spent caring for and researching cystinosis...more and more families becoming advocates for their own children and adults advocating for themselves. A real shift in not only the cystinosis community but also the greater community of rare disease has occurred over the past ten years that our family has been touched by this disease.

With these accomplishments noted and appreciated, The Cystinosis Research Network has made a concerted effort to search for the missing pieces. Which aspects of the community’s needs are not being met? In the past, this question has resulted in the idea to establish an endowed clinical fellowship at the National Institutes of Health under Dr. William Gahl to focus primarily on the care and research of patients with cystinosis. We were fortunate to have Dr. Galina Nesterova accept the position this Spring.

It was also noted this year that as our children are being diagnosed earlier, treated more effectively through adolescence and maturing into accomplished adults, there was no existing reference for families and professionals to utilize in guiding them through the journey of transition from pediatric and adult care. The Cystinosis Adult Care Initiative group has been working for the past several months to address this issue.

Families continue to express their most pressing need as the ability to connect with other cystinosis families. We look to expand the ways in which this can occur. We have increased our support for local family gatherings this year and have begun work on our biennial family conference to take place July 14-16, 2011 in San Francisco, California. We have added a link to the website where you can find information about other families in your area (http://www.cystinosis.org/families-in-your-area). CRN also continues to host our General Support and Teen Support Email lists as well as a Facebook Page and Cause – these avenues for connection are essential daily meeting places for many in our community. We view our role in facilitating the ability for the community to come together and share as one of our greatest and most vital accomplishments.

In this issue of The Cystinosis Advocate you’ll find more information about all of the above, as well as further updates from all of our standing committees, including:

**Finance** – CRN has completed our annual audit, which confirmed the financial health of our organization. Jeff Larimore has accepted the position of Treasurer, taking over from Brittney LeBeau, who stepped down from the position this summer after four years of dedicated service. CRN’s support comes from a wide range of individuals and companies, without whose generosity CRN would not be able to function.

**Research** – New and promising research is being funded by CRN. Look for updates in this newsletter on current progress. Announcements regarding newly funded research projects will be available soon.
Please check out the CRN website for the newly added Article Library, a bibliography of many critical research and reference articles about cystinosis for families and professionals.

This issue includes a summary of the results of the Cystinosis Patient Survey, conducted this summer in conjunction with Sigma Tau Pharmaceuticals. Thanks to everyone who participated, the information gathered has enabled Sigma Tau to prepare for the anticipated approval and launch of the cysteamine eye drops in the very near future.

Finally, the International Pediatric Nephrology Association meeting took place in August. CRN sponsored the concurrent Cystinosis Symposium which attracted hundreds of participants from around the world. Information from the event is included in this newsletter. The program book from the event can be accessed via the home page of the website at http://www.cystinosis.org/filemanager/file/CRNProgramBookFINAL.pdf.

**Family Support** – As mentioned, this issue contains information about the Cystinosis Adult Care Initiative, photos from this summer’s Family Gatherings, and other Family Support updates. The 2011 Family Conference is our greatest focus now. In consideration of the beautiful setting of this year’s conference in San Francisco and CRN’s efforts to assist in the community’s growth, our theme this year will be “Bridge to the Future”. We hope you will plan to join us next summer.

**Education and Awareness** – You will find information on this committee’s activities, including updates from cystinosis groups around the world, scholarship information, updates from our National Organization for Rare Diseases board member Marybeth Krummenacker and more.

Finally, I’d like to welcome our new board members, Colleen Hammond, Pam Woodward, Jessica Jondle, Jeff Larimore, Jenni Sexstone and Rich Jordan. All have already made significant contributions to CRN and the community and we thank them for their dedication and selflessness in their service on the CRN Board of Directors.

We remain dedicated to our mission of not only working to find improved treatments and an eventual cure for cystinosis, but also never losing sight of the importance of providing support to our community. Please consider becoming involved, whether it’s as a committee volunteer, fundraiser host or board member, we need your help and you can make a difference.

Warmest Regards,
Christy Greeley
President and Executive Director

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**Cystinosis Research Network Awards NIH Cystinosis Fellowship (cont.)**

Continued from Page 1

NHGRI, research into Fanconi’s syndrome and nephrocalcinosis in cystinosis patients and molecular testing of cystinosis patients. She intends to also maintain cystinosis databases, work to further educate The medical community about cystinosis and promote public awareness of the disease. Dr. Nesterova has committed to becoming a personally available resource for CRN, the medical community and families and individuals living with cystinosis. She can be contacted at nesterovag@mail.nih.gov with any questions or concerns regarding cystinosis diagnosis or treatment.

CRN gratefully acknowledges the Harry A. and Margaret D. Towsley Foundation and Eastside High School, Greenville, SC, for their generous financial support of the NIH Cystinosis Fellowship.

**CRN Family Conference Set for July 14-16, 2011 in San Francisco, California (cont.)**

Continued from Page 1

More information on the Hotel Nikko can be found on their website at www.hotelnikkosf.com. Ideally located in the very heart of the city, Hotel Nikko is a short walk from fine shops, theaters and restaurants at Union Square. We are so fortunate to meet in San Francisco next year, a beautiful city which will provide a wonderful backdrop for our community to gather.

Registration materials will be available early next year on the CRN website and will also be mailed to families. The Jordan and Jondle families have graciously agreed to host the event -- we thank them and the rest of the planning committee for committing to staging the conference. If you have any questions or suggestions for the conference, please feel free to contact the planning committee at info@cystinosis.org.

See you all in San Francisco!
CRN Education and Awareness Update

By Paula Shal
Vice-President, Education and Awareness

I have been writing the Cystinosis Research Network newsletter for the past five years, and it has been a unique vantage point to witness the progress CRN has made. I used to have to scour for submissions, and now there is more newsworthy information than we can fit in one issue!

There are exciting initiatives taking place in Research and Family Support as well as Education and Awareness. We are so proud that Marybeth Krummenacker was asked to be the Chair of the NORD Advocacy Committee. Anyone who knows Marybeth or has seen her speak knows that she will represent NORD well in this capacity, which benefits all rare disorders.

On page 20 is a list of the medical meetings CRN will exhibit at in 2011. We are always looking for volunteers to discuss cystinosis and exhibit at these meetings. At the time of publication Tahnie and Pam Woodward just returned from exhibiting at ASN in Denver, CO. ASN is the American Society of Nephrology and is primarily attended by adult nephrologists. As many of our children with cystinosis transition into adulthood, adult nephrologists are a very important group to educate. As an adult with cystinosis, Tahnie was a great representative at ASN.

By Paula Shal
Vice-President, Education and Awareness

CRN Family Support Update

By Cheri Friend
Vice President-Family Support

The CRN Family Support Committee supported our community by sponsoring a dinner at the Midwest regional family gathering which was held in Story City, Iowa July 23-25. The family gathering was hosted by the Hoffmann family (see write up from Doretta Hoffmann on page 21). CRN encourages our cystinosis families to hold regional gatherings especially during the off years when we do not hold our family conferences. If you are interested in hosting a family gathering in your area, please contact Cheri Friend at cfriend@cystinosis.org or (678) 546-4798.

The Family Support Committee also oversees the Adult Care Excellence Initiative that is currently in progress. The members of this focus group meet on a monthly basis to report on the progress of their sub-groups. The initiative is divided into 3 subgroups: 1) Clinical Practice Guidelines for Cystinosis – Pediatric through Adulthood, 2) Transition from Pediatric Care to Adult Care, 3) Identify Research and Treatment Needs to Improve the Quality of Life for Adults with Cystinosis. Each subgroup is diligently working towards the end goal of providing printed guidelines/materials related to each of these topics at the upcoming family conference in San Francisco.

We are proud to have awarded TWO sibling scholarships to Nikolaus Gard and Laurin Friend. See page 15 for more information about them. Applications for our 2011 scholarships for both siblings and individuals with cystinosis will be available in the first part of 2011.

And speaking of scholarships, one of my roles for the 2011 Family Conference is to lead the process of awarding scholarships that provide assistance for some families attending the conference. At CRN, we recognize times are tough, and we are committed to helping families attend as much as possible. Stay tuned for application information in early 2011 at www.cystinosis.org. Hope to see you in San Francisco!

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By Cheri Friend
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Marybeth Krummenacker Appointed Chair of NORD Advocacy Committee

The Cystinosis community is ONE community but we all come together with different voices and different opinions and backgrounds. The opportunity to be heard is just that --- an opportunity — to respect each other’s differences but speak with one voice. Cystinosis Research Network is fortunate to have Marybeth Krummenacker representing the cystinosis community on the Board of Directors of NORD (National Organization for Rare Disorders), which represents over 6000 rare disorders both in Washington D.C. and around the world. NORD speaks with one voice on behalf of the 30 million people living with rare disorders......of which cystinosis is one.

NORD Board of Directors President, Frank Sasinowski, recently asked Marybeth to chair the Advocacy Committee for NORD. With the advent of health care reform, there are changes happening almost daily. Whether we agree or disagree, it is important to pay attention to how it may affect our community. Marybeth has over twenty years experience as an advocate for both cystinosis and other organizations. This new position will allow her to be a “player at the table” regarding the issues relating to cystinosis and all rare diseases and how those issues will impact our lives every single day. As this newsletter goes to press, Marybeth will be traveling to Washington once again for the Board of Directors meeting as well as the Fall Corporate Council Meeting.

As the Advocacy Committee sets their agenda, Marybeth would like to stress that making contact with your local and national officials to voice your concern has never been more important.

European and U.S. Research Leaders Announce Collaborative Effort

Dr. Bill Gahl recently participated in a meeting in Iceland in which the European Commission and the NIH agreed to coordinate research funding. The article and photo below were reprinted with permission from NORD.

Following a meeting in Reykjavik, Iceland, the European Commission and the U.S. National Institutes of Health (NIH) have announced their intention to join forces on rare disease research. The two institutions plan to coordinate their research on rare diseases and to make major investments in this research field in the years to come.

The objective is to accelerate medical breakthroughs for people affected by rare diseases. Currently, although there are nearly 7,000 diseases recognized as rare in the U.S., only about 200 of them have specific treatments.

NORD President and CEO Peter L. Saltonstall and Sharon Terry, President of the Genetic Alliance, participated in the meeting in Iceland as representatives of the patient community in the U.S.

"We believe this type of international collaboration represents a major step forward in rare disease research," Mr. Saltonstall said. "NORD fully supports efforts to join hands across the ocean to improve the lives of patients and their families."

Dr. William Gahl (middle row, far left), Clinical Director of the National Human Genome Research Institute at the NIH, attended the European-USA Bilateral Workshop on Rare Diseases and Orphan Products meeting in Reykjavik, Iceland in October, 2010.
By Christy Greeley, Interim Treasurer

It is with the greatest respect, admiration and our deepest gratitude that CRN accepted the resignation as Treasurer from Brittney LeBeau in September. Brittney has worked tirelessly over the past four years to ensure that CRN's financial systems adhere to the highest level of scrutiny for a non-profit organization. During this time period CRN has grown from a small to a medium sized non-profit. To match that growth, all of our financial systems had to be updated, including transfer of all accounts and registration of CRN in the State of Illinois, adoption of the Quickbooks system, hiring of an accountant for taxes and bookkeeping services, and finally the incredibly time consuming process of yearly audits. All of these systems which Brittney is directly responsible for instituting ensure that CRN is completely financially transparent, compliant with all non-profit rules and regulations, and fiscally healthy for many years to come.

In addition to this work, Brittney has been responsible for all CRN budgeting, expense reporting, checks and deposits, and many other additional duties such as printing and mailing of the CRN newsletter. She has also held multiple fundraisers, raising over $100,000 for CRN. All of this while caring for her two beautiful children, Alexis and Evan, who both have cystinosis. She is a model of hard work, diligence and integrity. We have been blessed as a community for her contributions and we wish her all the best in her future as she goes back to school and focuses her full attention on her family. Brittney will remain on the CRN Board of Directors until her term ends in July 2011 to assist with transition issues. We welcome Jeff Larimore as our new Treasurer. Jenni Sexstone has also taken on a portion of the Treasurer role as well in her capacity as a new board member and member of the Finance Committee by overseeing the 2011 Conference Budget. Please help us to thank Brittney for her amazing and invaluable contributions to CRN and the cystinosis community!

This July, CRN’s annual audit for fiscal year 2009 was completed. In 2009, CRN received approximately $450,000 in total income with $375,000 in total expenses, giving an overall net income of $75,000 for the year. Nearly 70% of the total expenses went toward research. We are deeply grateful for the support of so many of the cystinosis families and their friends and colleagues. We would not be able to support the community without your help!

For the current fiscal year January through November 2010, CRN has received over $300,000 in donations, with many donations still expected the last month of the year. Expenses have kept on track with budget expectations. We anticipate meeting our fundraising goals for the year and look forward to a financially sound 2011.

Living with Cystinosis Survey

The Cystinosis Research Network requests your help to identify issues of concern in the cystinosis community, and to better understand how cystinosis affects the lives of patients and families. The Living with Cystinosis Survey addresses issues related to Living with Cystinosis and Loving Someone with Cystinosis.

The information collected from this survey will be used to design and support programs, services, and advocacy efforts on behalf of individuals with cystinosis. It will also be used in the Adult Care Initiative Project.

The results of this survey are anonymous and confidential. The survey may take 15-20 minutes to complete. The link to the survey is on the CRN website at www.cystinosis.org. Please note: This is a different survey than the Cystinosis Patient Survey that was open in July and August and had an emphasis on eye drop usage (see results from this survey on page 11.) WE TRULY APPRECIATE YOUR PARTICIPATION!
Raptor Pharmaceutical Corp DR Phase III Clinical Trial Started

Six US and French hospitals are recruiting cystinosis patients for a Phase 3 clinical trial of Raptor’s Cysteamine Bitartrate Delayed-release Capsules (DR Cysteamine). Below is the patient recruiting ad for the US sites.

**Phase 3 Study of Cysteamine Bitartrate Delayed-release Capsules (RP103)**

This multi-center research study will investigate a new cysteamine drug (RP103) for the potential twice a day treatment of cystinosis. RP103 will be compared to the existing four times a day treatment (Cystagon®),

The study will require the time commitment of 20 clinic visits over 10-11 weeks. Most of these clinic visits occur in clusters of 3-4 days in a row:

- **Screening Visit (Week 1)**
- **Run-in:** Weeks 2 and 3
  - take Cystagon®
  - 3 clinic visits during week 2; 1 clinic visit during week 3
- **Period 1:** Weeks 4, 5, 6
  - take RP103 or Cystagon®
  - 3 clinic visits during week 4; 3 clinic visits during week 6
- **Period 2:** Weeks 7, 8, 9
  - take the opposite of what was taken during Period 1
  - 3 clinic visits during week 7; 3 clinic visits during week 9
- **Follow-up Visit, 1 week later**
  - or the option to enter a longer study with RP103

There will be blood draws, overnight fasts and other evaluations. Travel and accommodations for participating in the study will be provided at no cost to the patient. Daily living expenses for a family member or guardian accompanying a minor will also be covered.

Eligible patients must be on a stable therapeutic dose of Cystagon®, be able to swallow Cystagon® capsules whole, not have received a kidney transplant and take all medications orally, not through a gastric tube. More information can be found at [www.clinicaltrials.gov](http://www.clinicaltrials.gov).

An extension study to determine the safety of long-term administration of Cysteamine Bitartrate Delayed-release Capsules is planned. Patients who complete this Phase 3 study will be offered the opportunity to be treated with Cysteamine Bitartrate Delayed-release Capsules until they are approved by the FDA or until Raptor Therapeutics withdraws its application with the FDA (for whatever reason).

If you are interested in participating and would like more information, please contact:

- Margo Kamel, working with Dr. Larry Greenbaum at Emory University in Atlanta, GA
  - Email: cystinosistrial@oz.ped.Emory.edu
  - Phone: 404-712-9923
- Heather Price, working with Dr. Craig Langman at Children’s Memorial Hospital in Chicago, IL
  - Email: hprice@childrensmemorial.org
  - Phone: 773-755-6368
- Xiaoxiao Gao, working with Dr. Minnie Sarwal at Stanford University Medical Center in Stanford, CA
  - Email: xiaoxiao@stanford.edu
  - Phone: 650-521-6072
CRN Research Update

By Elva Smith-Vice President-Research

Research studies for the improved treatment, and ultimately a cure, for cystinosis continue to be a vital part of the basic commitment of CRN to the cystinosis community. At this time, we have a total grant commitment of nearly $700,000.00. CRN remains most appreciative of the expertise and dedication of our grant recipients, and we look forward to expanding our support in promising new studies.

In the past six months, two of our research grants have reached conclusion:

Jess Thoene, MD, University of Michigan, Ann Arbor, "Tissue Repository for Cystinosis". Grant awarded: 30 August 2005, interrupted immediately by hurricane Katrina. Request for time extension, received 31 August 2007; request granted to "on-going". Total award: $26,206.00. Progress report received 22 November 2007 and FINAL report received 15 September 2010. Dr. Thoene’s payment for this grant is complete. The two reports can be found on the CRN web site www.cystinosis.org under Research/Research Updates.

Paul Goodyer, MD Montreal Children's Hospital, Montreal, Canada, and Rick Kaskel, MD Children's Hospital at Montefiore, Bronx, NY. "Proposal for a North American Cystinosis Research Platform". One year grant of $36,000.00 awarded for development. Payment has been provided and was equally divided between Drs. Kaskel and Goodyer. The Research Platform is currently in the planning stage and Drs. Kaskel and Goodyer are collaborating with other organizations in the interest of streamlining the registry projects currently in existence.

CRN’s Most recently awarded grant study:

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.

Studies that are on-going are as follows:

Elena Levtychenko, MD, PhD University Medical Center Nijmegen, The Netherlands, "Study of ATP metabolism in human cystinotic proximal tubular cells and in humans with cystinosis in vivo" Grant awarded: 11 September 2006 for 1 year, January 2007 through December 2007. Total award: $68,090.00. A 6 month Progress report received 10 July 2008. "Final report" received 5 February 2009. However, an email from Dr. Levtychenko indicated that the "In vivo part of this project is not finished yet due to the technical difficulties with the MRS apparatus, but we have good hope that the problem will be solved and this part of the study will be finished in 2009". CRN acknowledged receipt of this report, but has not accepted it as a "final" report. Final payment has not been provided as of 15 October 2010. An update from Dr. Levtychenko was received in September 2010 as follows: "Regarding the ATP study, in Leuven (and in Nijmegen) the liver coil is not operational yet, this is a major problem, which I can’t overcome, because I'm dependent of the technique which is not yet ready. Technical department still makes promises... I'm mailing them every two months. We have now a good lab in Leuven/Belgium, which works in close collaboration with Nijmegen, so for sure will apply for the new projects." Dr. Levtychenko is now associated with the Department of Pediatric Nephrology, University Hospital, Leuven, Belgium. Balance remaining on grant: $17,022.50

Continued on Page 9
CRN Research Update (Cont.)

Catherine Tuleu, PhD University of London, School of Pharmacy (other names on grant: Ken Nischal, Olufemi Rabiu, Rajnish Sekhri, Wm Van’t Hoff and Bola Lawal) “Development of cysteamine in situ gelling system for the topical treatment of corneal crystals in cystinosis”, Grant awarded January 2007 for 3 years, 1 March 2007 – February 2010. Total award: Pounds 103,000.00, approximately US$203,500.00 at that time. Extension of time requested due to Bola’s illness; granted 1-21-08. 1 year progress report rec’d July 2008. Balance remaining on grant: 67,400 pounds. Final report is still pending.

Leticia Belmont, M.D., Unidad de Genetica de la Nutrician, Instituto Nacional de Pediatria, Mexico. “Determination of Intraleucocitary 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. A presentation, citing CRN funding, was given at the IPNA meeting in NYC, 31 August 2010 and a progress report was received September 2010. Dr. Belmont stated in an email received 8 October 2010: “We consider it important to repeat the methodology with the new reagents and column to determine if these settings can achieve the objectives of the determination by HPLC and then compare it with the determination by tandem MS/MS that would be another project.” CRN has encouraged Dr. Belmont to conduct the repeat studies and submit a new proposal for the mass spectrometry comparison.

Hank J. Blom, M.D., M.M.C. Wamelink, and E. Levchenko, 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, or approximately $67,500.00 at that time. Initial payment was processed; interim report still pending.


Ewa Elenberg, MD, Texas Children’s Hospital, Houston, TX "Quality of Life in Cystinosis Patients", Grant awarded 25 September 2009 for 1 year (1 October 2009 to 30 September 2010). Total award: $21,000.00. Initial payment of $5,213.00 made on 12 April 2010. Balance remaining on grant: $15,750.00.

The third annual observance of Rare Disease Day U.S. will be February 28, 2011. Cystinosis Research Network will be a Rare Disease Day partner. Visit http://rarediseaseday.us/ to find out how you can be involved.
Over five years ago, the Cystinosis Research Network was given an opportunity. Dr. Frederick Kaskel asked us to be a major sponsor of the International Pediatric Nephrology meeting (IPNA) that was going to be held for the first time in New York City. CRN and its Board of Directors jumped at this opportunity.....even though we may not have been sure of what we were getting ourselves into! It was a commitment that we felt, as an organization, was one that we needed to make.....and so we did. Throughout the five years there were many telephone conference calls with key people involved in both setting an agenda and playing an active role as a sponsor. The opportunity finally came to fruition this past August. The 15th Congress of the International Pediatric Nephrology Association took place in Aug. 29-Sept. 2, with Dr. Kaskel leading CRN as the President of the Congress.

As part of our commitment to being a major sponsor of this historical meeting, on August 31st a half day session was held devoted to the lastest research in cystinosis from around the world. The afternoon began with a box lunch provided by CRN as well as a "poster session". Dr. Bill Gahl served as the moderator as well as organizer of the entire agenda. There was an opportunity for doctors from around the world to listen and learn from the world’s leading researchers and scientists on everything from the Mouse Models for Studying Cystinosis by Dr. Corinne Antignac, Newborn Screening by Dr. Silhoun Hahn, to the North American Cystinosis Research Network Registry by Dr. Paul Goodyer. The day was filled with information, timely topics, and Q&As after each speaker. CRN Board Member Marybeth Krummenacker was a speaker representing not only CRN but the National Organization for Rare Disorders (NORD), where she sits on the board. Marybeth spoke from her personal experiences and 20+ years of living with cystinosis. It was a productive day for CRN and the cystinosis community!

CRN hosted a dinner for the speakers the night before the meeting so that they could network and informally discuss cystinosis issues. CRN is honored to have had a once-in-a-lifetime chance to partner with IPNA. Thank you to Dr. Kaskel for giving us this opportunity.
Survey Sheds Light on Needs of Cystinosis Patients

As many of you know, throughout July and August, the Cystinosis Research Network (CRN) fielded the Cystinosis Patient Survey designed to help CRN and its partners better understand and address the unique needs of the cystinosis patient community.

CRN is happy to report that we received survey responses from more than 150 cystinosis patients in the United States. A special thanks to those of you who participated. The time you spent completing the survey questions will help us to identify new approaches for helping patients and their families face this rare disease.

Through the survey responses, we were able to identify various key learnings regarding those living with cystinosis in the United States and particularly, their experience with the cysteamine eye drops.

We are happy to share top-line results with you and hope you find the information enlightening.

CRN looks forward to working with its partners to determine how best to implement the knowledge gleaned from the Cystinosis Patient Survey. Please feel free to contact CRN with any questions regarding the survey project.

*The Cystinosis Patient Survey was made possible thanks to support from Sigma-Tau Pharmaceuticals, Inc.*

**Cystinosis Patient Survey Key Learnings**

Cystinosis patients and their families get information from a variety of resources, but primarily from their nephrologists and from patient advocacy groups.

![Chart showing where patients get cystinosis information and resources](chart)

Patients report that the most helpful resources at diagnosis include disease information, treatment information and meeting other cystinosis patients & families.

![Chart showing most helpful resources at diagnosis](chart)

*Continued on page 12*
Patients report wanting to have more information regarding doctors that treat cystinosis. Additionally, they indicate they would like new resources, access to online support groups and financial help for treatment.

About half of all cystinosis patients have received at least one kidney transplant. Almost all patients report having experienced crystal formation in their eyes.

More than half of the patients that have experienced crystal formation in their eyes use the cysteamine eye drops for treatment.

Patients that do use the eye drops tend to follow the recommended procedure for storing the drops in the refrigerator. However, some patients report not following the procedure.
CRN Welcomes Six New Board Members

Cystinosis Research Network added six new members to its Board of Directors in July. Two of the six are returning to the board after a short hiatus. All members bring an abundance of knowledge and experience and are sure to keep CRN headed towards our mission and vision. New board members include:

**Colleen Hammond**

Colleen and her husband Jack founded CRN in 1996. They live in Massachusetts. Colleen and Jack’s son, Shea (age 21), has cystinosis and has received a kidney from both of them. Colleen took a hiatus from CRN to obtain her nursing degree. She is currently leading the adult transitioning project for CRN. CRN is thrilled to have Colleen back on the board.

**Jessica Jondle**

Jessica is a 28-year-old with cystinosis and is a full time middle school teacher. She lives in California with her husband, Wayne, who just got out of the Marine Corp. Jessica previously worked in the publishing industry as a proofreader, writer, and, most recently, an editor. Jessica is interested in helping adolescents with cystinosis deal with self-image and coping issues. The Jondle family is one of the host families for the 2011 CRN Family Conference in San Francisco.

**Rich Jordan**

Rich lives in California with his wife, Mary, and their four children—Caitlin, Patrick, Joe and Conner. Joe has cystinosis. Rich is a Communications Team Leader at CoreNet Global WorkPlace. Rich earned a BS in Industrial Design from San Jose State University and an MBA in New Venture Management from California State University-Hayward - School of Business and Economics. The Jordan family is one of the host families for the 2011 CRN Family Conference in San Francisco.

**Jeff Larimore**

Jeff Larimore lives in South Carolina with his wife, Katie, and their four children—Jessica, Caroline, Daniel and Sarah. Sarah has cystinosis. Jeff is the CFO and COO of Southern Realty Development Corporation of Columbia and Arnold Construction Corp. He earned his BS in Accountancy from the University of Akron and is a Certified Public Accountant. Jeff has accepted the role of Treasurer for CRN, replacing Brittney LeBeau.

**Jennifer Sextone**

Jennifer lives in Ohio with her husband, Jim, and their two daughters, Jordan (age 6) and Josie (age 2, with cystinosis). She works as a Financial Analyst for F&W Media. Jenni received her BSBA with a double major in Accounting and Finance from Ohio Northern University and has her MBA from the University of Dayton. Jenni will be serving on the Finance Committee, and she is already assisting with the budget for the 2011 Family Conference in San Francisco.

**Pam Woodward**

Pam is returning to CRN after a short hiatus. Pam was the Vice President-Family Support for two terms. Her family also hosted the 2005 CRN Family Conference in Salt Lake City, Utah. Pam’s knowledge of CRN and family contacts will be an asset to the future of our organization.
Delayed-release Cystagon Studies at Emory University Bring Cystinosis Families Together

By Elva Smith and Frankie McGinnis

It’s not all about blood draws, EKGs, and new capsules to swallow. Raptor’s DR Cyta-teamine (RP 103) research study is now taking place in several locations in the United States. For the patients coming to Children’s Health Care of Atlanta at Emory University, there are some pretty special perks. The best is the camaraderie and fun the young folks are experiencing as they share time together at the clinic and at local fun events.

At the clinic, they learn that they are not alone in their cystinosis world. They discover friends, both old and new, who are also struggling with swallowing pills, gulping down gallons of water, making frequent trips to the bathroom, having to wear dark glasses in the bright sunlight, dealing with such things as cravings for certain food and ticklish tummies, and waking up nightly to take their life-saving medications. Hopefully, the latter will soon be a thing of the past as the 6-hour meds are replaced with the 12-hour time-release cystagon.

The folks coming to Emory have established a special bond. There’s Dr. Greenbaum, of course, with his bright bow-ties and juggling acts, as well as the wonderful, compassionate nurses who seem to have everlasting patience. And the kids themselves, along with their parents, siblings, and grandparents who have congealed into one big family unit. A lot of the thanks goes to Lorna Smith who has opened her home and vast back yard as a playground for nerf-dart battles, lego-building contests, and even an occasional quick-dip in the now very cold swimming pool.

The cystinosis families have individually enjoyed trips to the Georgia Aquarium, the Coca-Cola Expo, the Center for Puppetry Arts, and Stone Mountain. Group events have included an exciting dinner at Medieval Times with knights in shining armor, dancing horses, simulated sword fights and lancet thrusts. A big thank you for this event goes to Jonathan Rico. Other weekend excursions included trips to Six-Flags Over Georgia and the Gwinnett County Fair, and a Halloween Party!

A special dinner was held on Tuesday, October 19th at the Mexico City Gourmet restaurant. In attendance were the Smith, Rico and Friend families from Georgia, the Patterson family from North Carolina, the McGinnis family from South Carolina, the Binder and Thomas families from New York, the Reed family from Texas, and the Jones family from Florida. It was a very noisy but a most heart-warming event! The wait staff eventually decided to leave pitchers of water on the tables! A special thanks to Frankie McGinnis for making the arrangements as well as to Mason Reed’s grandparents, Frank and Rhonda Broce for their generosity when we all got the surprise that they had picked up the tab!

Cystinosis is not fun; but with such an awesome group sharing its challenges, much of the sting seems to go away. Along with a search for better treatments and ultimately a cure, that is a basic goal of the Cystinosis Research Network: unconditional support for the cystinosis family.
Two Recipients Awarded Sierra Woodward Sibling Scholarship

The Sierra Woodward Sibling Scholarship was awarded to two siblings of an individual with cystinosis. The 2010 recipients are Nikolaus Gard and Laurin Friend.

Nikolaus is the brother of Mikaela Gard, age 16, with cystinosis. He is extremely grateful and will use the scholarship to pursue a degree in computer science in hopes of becoming a computer programmer. He is currently attending Indiana State University in Terre Haute, Indiana.

Laurin Friend is currently in her junior year at Kennesaw State University. She is working towards her degree in early childhood education. Upon completion of her degree, she hopes to teach first or second grade. Working with children has always been Laurin’s passion and she has already logged well over 200 hours volunteering in local elementary school classrooms. Laurin is the daughter of Matt and Cheri Friend. She has a 16-year old sister, Morgan, who has cystinosis. Her other sister, Jennifer, donated a kidney to Morgan in December, 2003.

Cystinosis Research Network recognizes that the entire family is affected when someone in the family has cystinosis, and we are very proud of Nikolaus and Laurin. CRN offers scholarships for siblings and individuals with cystinosis. Recipients can be at any stage in their post-secondary education. Information for the 2011 scholarships will be at cystinosis.org/scholarships in early 2011.

Megan Morrill Racks up Equestrian Accomplishments

Megan Morrill, age 14, received a kidney from her mom in May, 2009 and has a newfound energy and confidence! Megan has competed in equestrian events for many years and recently received several awards for the 2010 horse show season.

There are 25 kids in Megan’s show division, and she competed in three events. There are shows all summer, and at the end of the summer the points are added for their “all around score.”

Megan jumps 2’3” and qualified out of this jump level (a big accomplishment.) She competed in a higher jump level and placed 4th out of 25.

She placed 3rd out of 25 in Ridden Hunter—judges judge on the horse and rider as a team as they jump fences.

And Megan placed 2nd out of 25 in Equitation—judges judge on the rider’s technique as she jumps fences.
Annie Patterson’s Make-A-Wish Journal

Annie Patterson’s daughter, Emily (age 11), has cystinosis. She was granted a Make-a-Wish trip in June that was over one year in the making. Annie documented their vacation.

Day # 1 (Friday) – written early Saturday morning:

Yesterday was crazy busy and basically a blur in my mind. We left the house at 7am and stopped in Sanford to eat. From there, we headed to RDU. While we were waiting, I noticed a group of Marines at a nearby gate. Colby had just told me a few days earlier that he thinks he wants to be a Marine when he grows up... so, I took him over and explained to the group that my 13-year-old son thinks he wants to be one of them. They each shook his hand and introduced themselves. Colby was impressed and said they all looked "intimidating." After the introductions, I told them that we appreciate everything they do. It was a moment that made me proud to be an American. I’m sure I can’t even begin to fathom the sacrifices these men & women (and their families) make on our behalf. Saying “thank you” was the least I could do.

I then made my way over to our gate and explained to the attendant that Emily was on a Make-a-Wish trip. We were given a boarding pass to get on the plane first. Once on board, the pilot came out and took Emily to the cockpit with him. She was amazed by how cramped it was and how many buttons he had to use to fly the plane. Throughout our flight, the pilot talked to Emily over the intercom. He pointed out landmarks, and as we were landing, he said “Now Emily, on the left side, we can see your ship, the Disney Wonder.” Once on the ground, he even came back on and said "...and I hope you have a wonderful time Ms. Emily." She loved the attention!

In Orlando, we headed to the tram towards the Disney Cruise Line Bus area. On the tram, Colby brought it to my attention that one of our local celebrities had been on the plane with us and was riding on the same tram as well. Sure enough, there was Greg Fischel (the weatherman from our local TV station). At this point, it was pouring rain in Orlando so, in order to strike up a conversation, I asked “Mr. Fischel, how long is this rain going to last?” Him and his family got a kick out of it and were especially nice once I told them we were there for Emily’s Make-a-Wish trip.

We boarded the Disney bus and headed toward Port Canaveral. Once there, it was hectic for the next few hours. Going through security was a breeze, but then you step on this big ole ship it’s like “OMG – where do we go first??!” We checked out our stateroom (and were delighted to find a cheese & fruit tray which our make-a-wish representative, Zoe, had left in our room...as well as a big ole stuffed “Captain Mickey Mouse” for Emily) and dropped off some of our carry-on bags. Before we left Orlando, tags were placed on our luggage to let the airlines know we were travelling on the cruise ship. Disney would be transferring our whoppin’ seven suitcases to our stateroom for us. It was nice to not have to wait for luggage claims in Orlando. Problem is, when our suitcases were delivered to the stateroom, they were soaked (victims of the above-mentioned rain which I’d asked Mr. Fischel about). So, we proceeded to hanging our clothes all over the balcony, in the room, on chairs, and anywhere else we could find space. We had a mandatory safety meeting on deck (unfortunately, I did not take any pictures of this... I’m certain we all looked quite fashionable with our bright orange life vests on). By the time the safety drill was over, it was time for dinner at Animator’s Palate. Don’t even ask me what I ate... cause I couldn’t begin to tell you what that stuff was...but it was pretty darn good. Ha! You can take the girl outta the country, but you can NOT take the country outta the girl.

Continued on page 17
Following dinner, everyone dispersed. Emily went to check out the Oceaneer kid’s club and lab. They issued me a pager so I could be paged when she was ready to be picked up. Colby managed to spend 20 bucks in the arcade within the first 20 minutes there. I walked around the ship and tried to figure out which end was which... still haven’t remembered what they call it but I know the “front” from the “back” and that’s all that matters.

We all finally made our way back to the room by about 10:30pm and started to settle in. Just when I thought it was time to sleep in, the princess (that’d be Emily) informed me she was “starving.” Room service delivered a good ol’ greasy pizza and French fries in the nick of time. She got her belly full and we all went into a vacation-induced coma. The gentle sway of the ship rocked me to sleep within moments of my head hitting the pillow.

Unlike anytime when I’m at home, I woke up bright & early this morning. I think knowing I don’t have to go to work had a lot to do with that. Before 6am, I was out on the balcony (oh, my bad, “verandah”) off our stateroom. Gorgeous, peaceful, relaxing... call it whatever you want, I LOVE it.

So, that brings us to the present moment. I’m still on the balcony, listening to Norah Jones on my laptop and we’re seeing land very close by. Nassau, we have arrived! Prepare yourselves! Todays agenda includes swimming with Dolphins, a little shopping, looking for Taylor Swift (she’s performing at the Atlantis Resort here tonight...so Emily is convinced we’ll run up with her), and drinking pina coladas (me, not the kids).

Life is great...and this is unlike anything I’ve ever experienced... I’m sure Emily would concur (if she wasn’t still piled up in the bed, snoring).

**Day 2**

I woke up bright and early and enjoyed the sun rise as we were pulling into Nassau. The kids enjoyed breakfast in bed...and then our day began. Nassau is HOT (and not like “pirate hot”... I’m talking more like INFERNO HOT). I knew Emily would have a hard time functioning in the heat so I packed a soaked towel in my bag for her and covered her with it while we were walking to the catamaran which took us to the Dolphin Encounter. While Colby thoroughly enjoyed the dolphin kissing and hugging him, Emily wasn’t real sure she wanted to interact that closely with such a large mammal. She chose to be more of a spectator...but she did rub it and said it felt like plastic. I think the dolphin needs Ritalin... he was pretty high-strung and squealed a lot... of course, maybe he’d just never kissed a southern girl. Hmm, who knows. Anyway... we were cramped for time on day #2. As soon as we finished with the big fish, it was time to head back to the ship where we each hurried through a shower and all-but-ran to make our 5:45pm dinner seating. Emily loved the attention of all the servers. When we returned from dinner, there was a note in our stateroom from Zoe inviting us to a “Special Meeting” on Sunday at 4:10pm. Emily instantly assumed it was Taylor Swift. Zoe had also sent us a nice plate of cookies to enjoy. Haha. Pizza @ 1 am... again.

**Day 3**

My favorite day of all! When I woke up this morning, we were already docked at Castaway Cay. After a quick breakfast, we disembarked. This place is friggin AWESOME! Make-a-Wish paid for us to rent tubes, floats, and snorkeling equipment for the day. We enjoyed a couple of hours swimming & snorkeling and took advantage of the barbecue hosted by Disney. For the first time during the trip, I actually knew what everything was that I was eating. Hehe. Emily struggled with her snorkeling equipment (afterall, it probably outweighs the child)... but she had a blast splashing in the water and trying to catch fish in her sand bucket. We reluctantly headed back to the ship in time for the meeting Emily had been invited to. There, she was able to meet the captain of the ship (Cpt. Guss) and the owners (Mickey and Minnie Mouse J ) . She was all smiles and felt like a true diva. I was glad to see she wasn’t disappointed about Taylor not being there. We left the meeting and headed to dinner where Emily once again soaked in the attention of the servers. She’s a ham... and I can totally see that it comes natural for her. Following dinner, we headed to the movie theatre for the premiere showing of “Toy Story 3” in 3D. On the way, we ran into Snow White on the staircase. She graciously moved aside and said...
“hello princess” to Emily. You can only imagine the look on Emily’s face... priceless! The movie was hilarious and the kids were all smiles. At 11pm, we sat out on our verandah and enjoyed fireworks. Disney is the only cruise line that is allowed to set off fireworks at sea. You haven’t seen fireworks til you’ve seen them in the middle of the ocean :0) At 1am, Emily and I had our nightly room service delivery of pepperoni pizza. I almost think they expected our order to come in... cause they had it to the room within minutes.

**Day 4**

Today was our “sea day” with no ports of call. The kids slept in until about 10am and enjoyed breakfast in bed (they’re going to really miss that luxury when we get home). Emily and I caught a show at 1pm called “Disney Dreams.” It had Tinker Bell in it, so she completely adored it! The rest of the day was pretty lazy... just coming & going around the ship... doing whatever we wanted to. Emily chose to see Toy Story again and Colby and I snuck in a couple games of Bingo (and I was called on stage and asked to do a River Dance, which I politely declined). We also managed to spend some money in the ship stores where Emily purchased a cute little stuffed baby elephant. I would say it is a baby Dumbo, but Emily says that is a hurtful word, and she has named it “Ellie” (get it – Ellie – ELEphant ). Tonight, she said goodbye to Ricardo and the other waiters whose names I can’t pronounce, spell, or even guess at. Following dinner, we discovered that most folks were in their staterooms packing up for departure. This left the pools nearly empty (up until this point, they’d been so packed that Emily hadn’t wanted to swim). Decked out in her bathing suit and goggles, she had a great time splashing around and watching "Planet Earth” on the huge screen outside. For the first time during the trip, I was able to just chill-out for awhile. Back in the room later that night, Emily told me “well mama, I think you could say that I really know how to make a wish.” I asked her if her wish had come true, and she replied with a smile “yes.” We were rocked to sleep one last time by the sway of the waves.

**Day 5**

Yikes! We were required to be out of our room by 8am! I was none-too-happy when I woke up this morning and saw Port Canaveral outside my window (a girl can get use to waking up to crystal blue water every morning). Emily was NOT a happy camper about the early wake-up call either. We did manage to get her to breakfast by 7 where she picked at her food and looked all-together pretty miserable. Haha. She gets it (non-morning-person-ism) from her mother. We said our final goodbyes and headed to the airport. There, we were able to sneak in a quick visit with one of our favorite people – Whitney Glaize. Seeing Whit’s beautiful smile calmed my nerves and put Emily in a much better mood. Our flight was uneventful, and we made excellent timing getting home. Once here, Emily went straight to bed and slept until I woke her up for dinner at 8pm. She’s exhausted (we ALL are), and I must admit that I feel a little depressed tonight. This trip has been over a year in the making. and, well, it’s just sad to me that it’s over now. I will forever be grateful to Make-a-Wish, Disney, and all the wonderful people who came together to make this such a special time for Emily. Her spirits are higher than imaginable right now. For complete strangers to open their hearts to your child, that’s something that words just can’t describe. It is humbling, to say the least.

Facing challenges, be them what they may, can cause a person to lose hope...to feel trapped...to think that everything is pre-determined, and there is no use in wishing for change.

Make a Wish proved to Emily that wishes really can come true.
Sisters and Best Friends Who Share Everything (Including Cystinosis!)

By Ashley Haynes

Our story began in March, 2003 when our family welcomed Lily Grace, a beautiful and healthy baby girl! For the first year of her life, my husband and I read the child development books and compared Lily’s progress weekly. We were so proud of each accomplishment! By her 1st birthday, we began getting concerned about some delays in her growth and development and by her 18 month check-up, we didn’t even look at the books. We began visiting specialists, therapists, getting tests and labs done. Finally, she was diagnosed with failure to thrive – a diagnosis that broke our hearts! A little after her 2nd birthday, a new specialist looked at some labs done by a previous specialist and discovered some abnormalities – he sent us directly to a pediatric nephrologist. We were relieved to finally have a diagnosis-something called Bartters Syndrome. The doctor prescribed some supplements which seemed to be helping. Lily began to grow again. She had more energy, and, best of all, she wasn’t vomiting daily!

In June 2006, when Lily was 3, our family received another gift; her sister, Maggie Jo. Maggie was another beautiful and healthy baby! Lily’s good health continued and Maggie’s too! In June 2007, our nephrologist broke the news to us that she had noticed something when looking at Lily’s labs. There were some changes indicating that Lily might have a disease called Cystinosis and that she wanted us to have Lily tested. We went straight to the hospital and had this done. Then we went home and looked it up on the internet! We were not happy with what we saw, so we spent the next month waiting for the results and hoping it wasn’t Cystinosis. Unfortunately, when the doctor called back, she said that it was Cystinosis and that now we needed to take Maggie and get her tested. Because Maggie had been so healthy, we tried to convince ourselves that she wouldn’t have it. When we got the call telling us that Maggie had it too, we were devastated. We went through three weeks of sheer frustration, trying to get into the routine of giving medication that tastes horrible around the clock! But our family worked together and within a few months, we had the hang of it! Bit by bit, things got easier, the medicine mixing and preparing, the getting up in the middle of the night, the extra expense! Within a year, Lily was taking all pills and when Maggie turned 3, she began taking all pills! This helped us out tremendously because now we don’t spend time mixing and drawing up syringes full of meds!

This past summer we were told that Lily needs a kidney transplant soon, and we have started making preparations for it. If everything goes as planned, she will be getting a transplant from me, her mom. We worried about talking to her about this because this would be the first time that she would really realize how different she was from the other kids. We worried, when we went to the transplant evaluation, that she would listen to all that was said and become sad or upset. Well, we didn’t need to worry! She skipped around that hospital and smiled at everyone. She did math problems and played with her princesses. She didn’t let that hospital scare her one little bit!

Lily, now 7 years old, and Maggie, now 4 years old, do everything together! They play in the yard or in their playroom for hours. They giggle, make up songs, dance, play school, play babies, draw, color, and, of course, fight! If one of them stays home while the other goes out somewhere, the one at home seems lost and lonely until the other one gets back. They are the best of friends! Lily Grace and Maggie Jo Haynes share everything: a family, a bedroom, toys, clothes, and a scary lifelong disease. And they know they are truly blessed to have each other!
Upcoming Family and Medical Events

March 7-9, 2011
Genetic Diseases of Children
Advancing Research and Care Conference
New York, NY

April 30-May 3, 2011
ASPN Annual Meeting
Denver, CO

June 23-25, 2011
2011 Genetic Alliance Annual Conference
Bethesda, MD

July 14-16, 2011
2011 CRN Family Conference
San Francisco, CA

November 8-13, 2011
ASN Renal Week
Philadelphia, PA

Frankie McGinnis Represents CRN at 2010 Genetic Alliance Conference

By Frankie McGinnis, CRN Board of Directors

Invisible in Everyday Life

On July 15-18th I had the pleasure of attending the 2010 Genetic Alliance Annual Conference in Washington, DC. The theme of this year’s conference was “Advancing Novel Partnerships,” which is Genetic Alliance’s mission as an organization bringing together numerous advocacy groups, researchers, policymakers, health professionals and industry representatives. There were many groups present and as always, more information than one person can absorb.

I attended a session called “Strategies for Sustainability and Success,” which was facilitated by Lisa Wise, COO of Genetic Alliance. This all-day discussion looked at the life cycle of an organization and gave the participants a chance to analyze where his/her organization was in the life cycle and identify ways to sustain viability. As I listened to other groups and their struggles, I realized just how far Cystinosis Research Network has come and how we have remained true to our initial vision. Our goal has been to continue searching for better treatments and to look at the affects of the disease on the entire family. We are in line with the advice from genetic counselors and physicians. They encourage patient groups to move away from the use of “cure” and more towards making disorder “invisible in everyday life”. Genetic defects are not something that will likely be fixed in our lifetimes. They are part of our genetic code, and organizations should look at the realities and focus on the best treatments which allow our populations to live the fullest life possible. False hope and empty promises can be a dangerous and deadly problem if a population is waiting for a miracle that most likely will not come. As a board member of CRN since its inception, I am proud of the change I have seen in our researchers and our population. Our researchers are embracing the longer lives our patients are experiencing, and our population is participating in research across the US and are objectively reacting to changes. I realized we are an “old” group now in the life cycle of advocacy groups, and I am satisfied with the adult we have become.

The conference also discussed volunteerism for advocacy groups. Often, a few members of the community for a disorder volunteer much of their time when in fact, the entire community is affected by the disorder. One idea which was presented is that if you have an hour to donate in lieu of a month or a greater time commitment or a financial donation, that hour has value. I hope CRN can use this idea and bring a more multi family approach to many of our existing committees. We all have something to contribute, and each family needs to own this group as its own. We are all in this together, and we are only as strong as our weakest link. Participation is vital to sustaining our vision of keeping our population healthy, our families together, and our organization moving forward.

Overall, CRN’s attendance and representation at the Genetic Alliance conference not only keeps cystinosis front of mind for those advocating for rare diseases, but also keeps us up to date as an organization on non-profit best practices and ways to better serve our families.
2010 Midwest Cystinosis Regional Family Gathering

By Doretta Hoffman

The Midwest family gathering was held on Friday, July 23 thru Sunday, July 25 in Story City, Iowa with Don, Doretta, Jon, and Andrew Hoffmann as the host family. We had nine families in attendance, which included eight children/adults with cystinosis. The Hoffmann’s welcomed the families with a welcome package and a gift to remind everyone to be an organ donor. On Friday night everyone was at the Hoffmann’s house for a meal of grilled hamburgers and brats. There were yard games to be played and a great game of ping pong. Later in the evening we watched a DVD that Jon Hoffmann had put together of the past 20 years of the Midwest cystinosis families and past conferences. It brought back many memories of past gatherings and how our cystinosis children have grown up to become adults.

Saturday morning a few people in the group went for a swim in the pool and then shopping at the local mall. At noon there was pizza delivered to the motel provided by the Cystinosis Research Network. The Midwest group would like to thank CRN for their support. After lunch the annual family and group pictures were taken. We were then off to a ride on the 1913 Herschell-Spillman Antique Carousel where Terry Tschannen and Eric Gard decided to fit right in the Norwegian heritage and purchased the Norwegian Viking hats! After the great carousel rides it was off to the bowling alley for a game of "Crazy Bowling". Before we started bowling, we displayed the traveling trophy to be awarded to the winner and to take home with them until next year. After the "Crazy Bowling" game was over Jon Hoffmann presented the award. He said that since this was "Crazy Bowling" there was a twist and the award would be presented to the person with the lowest score, not the highest! The trophy was awarded to Mikaela Gard.

After bowling we headed back to the motel to meet the Ridgeway family who are new to our group this year. After introducing ourselves it was back to the Hoffmann’s for a meal and more of the competitive ping pong and yard games. Later in the evening we had a campfire and sat around the fire and visited about where our children are in the cystinosis journey. Just two weeks prior to our gathering, we lost one of our own cystinosis adults, Keri Hohl. Since the Hohl family was able to come to our gathering, the Midwest families presented them with a concrete memorial garden plaque in memory of Keri. We were all able to help them with the healing process in the loss of Keri. Please remember to keep the Hohl family in your prayers in the days ahead.

The weekend was wrapped up with breakfast and frozen fruit cups at the motel and us all saying our goodbyes till we meet again. Robert and Sylvia Douglas family will be the host family for 2011 gathering.
The 6th International Cystinosis Conference was held in Lignano Sabbiadoro, Italy from September 23-26, 2010. The conference was held in a very nice complex that was located just minutes from the beautiful Adriatic Sea. The event included patients, families, and doctors from all over the world including Europe, USA, South Africa, Australia and more. The latest information on cystinosis research and treatments was presented in this conference. The highlight of this conference was a very special tribute to Dr. Jerry Schneider for his lifetime commitment of working on cystinosis. The tribute included an awesome slideshow that was created and presented by Melissa Scott of Australia. It ended with Dr. Schneider and his wife receiving a huge farewell cake and an enormous thank you from all.
Cystinosis Foundation Ireland Update

By Mick Swift

Cystinosis Foundation Ireland has had another busy and fulfilling year. Fundraising has become more challenging for all of us, but our great team of supporters continue to work hard to support our research efforts.

These events support our five research projects:

Professor Don Cairns and his team at Robert Gordon University, Aberdeen: “Design and biological evaluation of novel compounds for the treatment of cystinosis.”

Dr. Patrick Harrison & his team at University College Cork: "Development of an in vivo gene repair kit: towards a cure for cystinosis "

Professor Philip Newsholme & his team, University College Dublin, Project 1: “Why do cells die in Fanconi syndrome?”

Professor Philip Newsholme & his team, University College Dublin, Project 2: “Investigation into pancreatic dysfunction and muscle myopathy”

Dr. Minnie Sarwal and Dr. Poonam Sansanwal at Stanford University, California: “Investigation of the mechanism that can prevent renal injury in Cystinosis”

We learned of a new case of Cystinosis, a little boy in Northern Ireland whose family is already very active in fundraising and took part in a relay full marathon in Belfast this May.

Our fund-raising efforts have been helped by marathon and half marathon runners supporting us, a group of Dublin taxi drivers who held a five-a-side soccer tournament, a sponsored shave and now we are selling our first calendar (available online at www.cystinosis.ie).

We entered a national competition for community groups and came second in our area, winning €3,000 which went straight to cystinosis research. We will be entering again in 2011 and invite all our international friends to vote online.

Details will be on our website or on our Facebook page – why not join us! “Facebook.com/cystinosis.ireland”

Mick Swift, our Chairman, Anne Marie O’Dowd, Des Hennelly, Sue Maguire, and Andy Maguire all attended the Sixth International Cystinosis Conference in Italy in September. It was a great event, well done to all involved.

We were really delighted that six researchers from Ireland attended the conference providing a great opportunity to meet with other scientists and also to get a greater awareness of Cystinosis. The researchers presented at the poster sessions which included Professor Philip Newsholme and Rodolfo Sumayao from U.C.D., Dr. Patrick Harrison, Katrin Kaschig, Ciaran Lee and Jennifer Hollywood from U.C.C. and Professor Don Cairns and Barbara Buchan from R.G.U., Aberdeen.

Looking forward to seeing you all in San Francisco next summer. Please visit our website for regular updates, www.cystinosis.ie

Participants at the 15th Congress of the International Pediatric Nephrology Association in New York City included Dr. Jess Thoene (far left) and Dr. Bill Gahl (far right). CRN was a major sponsor of the event.
Mexican Association of Cystinosis Announces 2011 Board

Victor Gomez-President

My name is Victor Gomez. Most of you know me well. I am 28 years old with cystinosis. I founded the cystinosis group in Mexico City, and we have been changing cystinosis history in Mexico and Latin America for eight years. I accepted a second term as President because I am really interested in helping those in need, especially in my country and its surroundings. In Mexico we have some difficult issues. We don’t have a laboratory that makes cystagon. Our economy is poor, and we just don’t have doctors who are interested in cystinosis.

We are slowly making progress, but it hasn’t been easy. We will continue to work hard to make more progress. One of the most important events in my life is coming soon—I am getting married. This motivates me to continue to work with others to find a cure for cystinosis.

Julio Alberto Lopez Hernandez-Treasurer

My name is Julio A. Lopez, I am 36 and have worked in Advertising and am currently a filmmaker. I support my beautiful family, my wife and 2 sons. I became a board member for the Mexican Association of Cystinosis because my younger son Gael was diagnosed cystinosis at the age of 4. It took a long time for us to receive a diagnosis. He had a lot of lab tests and hospitalizations and saw many doctors. We have really important work to do to fight this disease, and we must help get kids diagnosed earlier. It is hard to find support, so we must take the initiative—share information and get doctors involved. We need to work to find improved treatments.

I have to point out that thanks to Mexican Association of Cystinosis my son Gael is on cystagon treatment and has medical care.

Graciela Niquet-Secretary

I am Graciela. My youngest son Josan has cystinosis and like everyone, we had a really hard time before he was diagnosed. We had no idea what was happening with him. He was diagnosed at age 5 and has had two kidney transplants. Currently he is doing okay.

Through Cystinosis, I met several people, including Leticia Belmont MD and Victor Gomez. I co-founded Mexican Association of Cystinosis in 2002. We have been working and improving over the past eight years. I decided to be part of the board because we have not yet finished our goal of helping cystinosis patients live a healthy life. I believe moral support is very important, too. That is why I am here.

From my heart I appreciate all patients that God has given me the chance to meet, such as: Miriam, Gerardo, Victor, Lupita, Irais, Angel, Carlitos, Silvia, Linaloe, Chuchito, Miguelito and my lovely Josan. A couple of them passed away, but their life and courage will remain in my heart.
Cystinosis Foundation UK Funds New Equipment for Cystinosis Research

By John Terry

The Cystinosis Foundation UK chairperson, Roy Forsyth, recently visited Sunderland University to present new equipment funded by a donation from the UK Foundation.

As part of our ongoing work with Sunderland University into the development of prodrug therapy for cystinosis, we were pleased to supply the project with a high performance liquid chromatograph.

This is used to analyse the compounds that have been produced and is a dedicated unit for the project. It will ensure that high quality, reliable results are achieved efficiently. Funding for the machine was allocated at the start of the research but it is only now that detailed analysis of the compounds has started that the University requested the funds.

Victor Gets Married!

Victor Gomez, founder & President of the Mexican Association of Cystinosis was married November 13, 2010 in Morelos Mexico.

Victor met his wife 20 years ago. Both were living in the same neighborhood, and their families used to go out and have coffee and dinner. They share the same religion.

By the year 2000 Maryan and Victor became good friends. They went out often to the movies, parties, and hung out with the same friends. They talked to each other about their feelings and other relationships. Both enjoy dancing more than anything.

By 2008, they were calling each other more. Conversations were different and longer, sometimes one to two hours. Victor started to have feelings for Maryan and gave her flowers one day, but it was in front of many people when going out to the movies.

In 2011 Mexican Association of Cystinosis will be working to include cystagon treatment through government medical service (social security) research projects. In addition, they will host the 6th Medical Symposium and collaborate with other organizations regarding rare diseases.

Stay tuned at www.cystinosismexico.com

Finally, he told her he loved her. She refused to listen a couple times, but by April, 2009, they were committed to one another.

The next year in April, 2010, victor took Maryan to Washington DC and proposed in front of the Washington Monument.
CRN Development Update

By Jen Wyman, Vice President, Development

Without continual growth and progress, such words as improvement, achievement, and success have no meaning. ~Ben Franklin

Growing pains are symptoms that an organization needs to make a transition and transitioning is exactly what the Cystinosis Research Network is doing. We have been working with a Development Consultant, Jose Hernandez, who has been an incredible resource for us. He is transitioning us into a more efficient, well equipped, well oiled, machine; outlining important areas and focusing on what works. Together we are making sense of our goals for expansion as an organization and creating a development plan that will grow with us as the years go on. The Cystinosis Research Network is unfolding and blossoming. It has been an exciting process, watching what we are doing right turn into something we can do better!

2010 fundraising has been successful thus far with approximately $300,000 raised this year. It is because of the amazing people behind the events, held on behalf of CRN, that we continue to raise significant funds. Thank you to everyone who was able to host a sporting event or write a letter campaign. It’s never easy to reach a hand out for a donation, but time and time again families within CRN do it and are rewarded with the generosity of so many who are touched by our stories. A special thanks goes out to the Woodward Family who hosted, once again, the C.H. Robinson Golf Outing and to John Maccarone and the Long Island Plumber’s Association for their efforts on two very fabulous and successful golf outings. Together they raised $85,000 for CRN.

CRN is a non profit organization that has a mission. It is dedicated to supporting research, providing family assistance and educating the public and medical communities about cystinosis. But it is more than that. It is a life support system for those families affected by this disease. It brings us knowledge, guidance, support and HOPE for a better tomorrow. Unfortunately CRN cannot sustain on HOPE alone; it also takes many dollars to keep an organization moving forward. CRN depends on its members and their fundraising efforts to support our mission. Our hope is that some of you will join us and venture into the world of fundraising. Anyone who has held an event knows that it is more than just raising money for a cause near and dear to your heart. It is also a heartwarming, gratitude filled, enlightening experience. Whether you raise $100 or $100,000 makes no difference. It will change you forever.

Please visit the website www.cystinosis.org. The fundraising tab has many ideas and samples for you to use as a resource in planning your own event. And as always please feel free to contact me at jenniwyman@att.net.

Never doubt that a small group of thoughtful, committed citizens can change the world; indeed, it’s the only thing that ever has. ~ Margaret Mead
# 2010 Fundraising Calendar

<table>
<thead>
<tr>
<th>Date</th>
<th>Event</th>
<th>Location</th>
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<tbody>
<tr>
<td>January 8</td>
<td>4th Annual Shoot for the Cure</td>
<td>Whitesboro High School</td>
<td>Mason Reed</td>
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<td>Whitesboro, TX</td>
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<td>January 30</td>
<td>Los Galos Hoedown BBQ</td>
<td>San Antonio, TX</td>
<td>Jon Ben and Ava Shepperd</td>
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<td>February 15</td>
<td>Eastland High School Cystinosis Awareness Week</td>
<td>Eastland High School</td>
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<td>Lanark, IL</td>
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<td>February 20</td>
<td>4th Annual Cause-2-Bowl</td>
<td>Bloomfield Hills, MI</td>
<td>Kacy Wyman</td>
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<tr>
<td>February 20</td>
<td>Texas Hold Em’</td>
<td>Vega, TX</td>
<td>Mason Reed</td>
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<td>March 17</td>
<td>Jack Greeley’s Birthday Fundraising Letter</td>
<td>Lincolnshire, IL</td>
<td>Jack Greeley</td>
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<td>April</td>
<td>Stilke Letter Campaign</td>
<td>Milwaukee, WI</td>
<td>Mason and Livia Stilke</td>
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<td>May 1</td>
<td>abeXChallenge</td>
<td>Cantrall, IL</td>
<td>Kenadee Julian</td>
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<td>May</td>
<td>5th Annual Golf Tournament</td>
<td>Berthoud, CO</td>
<td>Nevaeh Stanford</td>
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<td>May 2</td>
<td>4th Annual 5K Fun Run/Walk</td>
<td>Bloomfield Hills, MI</td>
<td>Kacy Wyman</td>
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<tr>
<td>June 12</td>
<td>4th Annual C.H. Robinson Golf Tournament</td>
<td>Lehi, UT</td>
<td>Tahnie Woodward</td>
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<td>June 13</td>
<td>Strike Out Cystinosis with the Cleveland Indians</td>
<td>Cleveland, OH</td>
<td>Jake Krahe</td>
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<td>June 15</td>
<td>Larimore Letter Campaign</td>
<td>Columbia, SC</td>
<td>Sarah Larimore</td>
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<td>July</td>
<td>Let’s Strike Out Cystinosis</td>
<td>Independence, OH</td>
<td>Victor Gardner</td>
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<td>August 23</td>
<td>Guest Bartending Event</td>
<td>Hinsdale, IL</td>
<td>Alexis and Evan LeBeau</td>
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<td>September 20</td>
<td>Plumbing Contractors Association of Long Island Golf Outing</td>
<td>Bay Shore, NY</td>
<td>The Maccarone Family</td>
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<td>October 1</td>
<td>Sextone Bowling Event</td>
<td>Hamilton, OH</td>
<td>Josie Sextone</td>
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<td>October 9</td>
<td>Hassan/Riffle Wedding</td>
<td>St. Leonard, MD</td>
<td>Austin Riffle</td>
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<td>October 17</td>
<td>Kacy Wyman Fundraising Letter</td>
<td>Bloomfield Hills, MI</td>
<td>Kacy Wyman</td>
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<td>October 23</td>
<td>Guest Bartending Event</td>
<td>Downer’s Grove, IL</td>
<td>Alexis and Evan LeBeau</td>
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<td>November 4</td>
<td>Senior Tennis Mixer</td>
<td>Dearborn, MI</td>
<td>Kacy Wyman</td>
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<td>November</td>
<td>Krahe Letter Campaign</td>
<td>Broadview Heights, OH</td>
<td>Jake Krahe</td>
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<tr>
<td>December</td>
<td>LeBeau Letter Campaign</td>
<td>Aurora, IL</td>
<td>Alexis and Evan LeBeau</td>
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Letter Campaigns: An Easy Way to Make a Big Difference

Dear CRN Families and Friends,

My name is Katie Larimore and I have happily accepted the challenge of coordinating Letter Campaigns for the Cystinosis Research Network. I live in Columbia, South Carolina with my wonderful husband Jeff and our four amazing kids. Our youngest daughter Sarah was diagnosed with cystinosis in October of 2006. She is now a tiny ball of energy at 5 years old. As a family we battle cystinosis every single day, and we hope every single day that it will be a healthy day for Sarah.

This past June 22 on Sarah’s 5th birthday, we sent out 100 copies of the most honest, terrifying and important letter that we have ever written. Our hearts were put on that paper as we shared Sarah’s story with our friends, family and associates. We wanted to explain the desperate need for research while at the same time giving back to the CRN for being our lifeline. Our daughter is alive today in large part because of the advice and contacts provided by the CRN. Our gratitude is immeasurable. We set a modest financial goal of $2,000. We not only exceeded our goal but gained more than we ever imagined. The generosity of friends and the warm words of love and support were overwhelming. We raised awareness of cystinosis in our community higher than we ever dreamed because many of those 100 original recipients forwarded our letter to their business associates, medical contacts and even their email contact lists. Based on our success we have committed to writing a Birthday Letter for Sarah every year until a cure is found.

Please consider joining Jeff and me and many other families coping with cystinosis by committing to a letter campaign in honor of your child and benefiting the CRN. I ask that you do one letter. I would wager that your personal rewards will be as great as the financial help you would be giving the organization that gives so much to all of us. I know what I am asking is not an easy task. I promise to help in any way. I am always available for your questions or just to discuss possibilities. Do not hesitate to contact me directly. There are past letters posted on the CRN website including our own. Please visit these for inspiration and to remember that we are all in this battle together and together we WILL find a cure. Until a cure is found it is vital that we all participate in spreading awareness and raising funds. The Cystinosis Research Network needs your help as much as you need the CRN.

Most Sincerely,
Katie Larimore
Klarimore112@sc.rr.com
803-873-2139

The Walgreen’s store where Jan Julian is a pharmacist sponsors “Jeans Day” for charity. In May, the store selected the Cystinosis Research Network in honor of Dan and Jan Julian’s daughter, Kenadee. Employees paid $3.00 to wear their jeans to work. In all, $689.00 was raised for CRN!
Krahe Family Shows Appreciation Through Fundraising

By Amy Krahe

In April of 2008 our son Jake was diagnosed with cystinosis. After spending weeks in the hospital Jake came home with an entirely new routine. He needed medications around the clock and a feeding tube 24 hours a day.

Over the past two years we’ve watched him endure countless blood draws, numerous doctor visits, ultrasounds, surgeries, x-rays and of course, the dozens of medications he takes every day. Until recent, Jake struggled with constant nausea and vomiting. Exhausted, we turned to the families of the Cystinosis Research Network(CRN).

Here, at CRN, we were greeted with an abundance of encouragement, direction and information. In July 2009 we attended our first Family Conference. We were introduced to families and medical experts, both offering an array of invaluable information. It was the first time we stood in a room with families that truly understand our heartache, our fight and our inspiration.

Today Jake is stable, gaining weight and beginning to grow! He is strong, resilient and joyful. He inspires us every day. He is a leader and, without a doubt, a true gift. He teaches us that we are strong and reminds us "Life isn’t about waiting for the storm to pass. It’s about learning to dance in the rain.”-anonymous

In appreciation of the families and physicians involved with CRN and to give Jake hope for a long and healthy future we decided to send our first letter campaign entitled Surviving Cystinosis in November 2009. We raised over $6,100 for CRN!

In June of 2010 we then decided to organize our first fundraising event. In cooperation with the Cleveland Indians, we hosted Strike Out Cystinosis, a Family Fun Day at the Cleveland Indians baseball game. We sold 216 tickets and raised over $4,300!

Later this summer Carter Lumber sponsored a Ball Drop for Cystinosis at the Medina County Home Builders Association Golf Outing. The Ball Drop winner Dan Clement, of Clement Construction Inc., also won the 50/50 raffle and donated 100% of his winnings to CRN. All in all the total donation to CRN was $1040!

With each event we find ourselves truly blessed. We realize that Cystinosis is rare, and everyone who participates does so in support of our family. Maybe they know my husband, my sister-in-law or maybe they are a friend of a friend. In some way they are here to support Jake. We are incredibly grateful!
The Cystinosis Advocate

C.H. Robinson Annual Golf Tournament Raises $55,000 for CRN

By Tahnie Woodward

On a cold and rainy Saturday in June, 2010, the 4th annual C.H. Robinson Golf Tournament to benefit CRN in honor of Tahnie Woodward, took place at Thanksgiving Point in Lehi, Utah. The little miracle girl, Sookie, (Rory and Tahnie’s daughter) was the special guest of honor during the luncheon. Everyone cheered as Rory held her up for all to see. An exciting raffle was held in order to increase additional revenue. This was made possible with the help of generous local companies donating wonderful products. It was a successful day despite the uncooperative weather. The event raised $55,000 for CRN! The 5th annual tournament is already in place for June 11th, 2011!

Tahnie Woodward and Sookie

Plumbing Contractors of Long Island Raise $50,000 for CRN at their Annual Golf Outing

By Marybeth Krummenacker

The Cystinosis Research Network was the beneficiary, for the second year in a row, of the 15th Annual Plumbing Contractors Association of Long Island Golf Outing, held on September 20, 2010. The outing was held on a picture perfect day for golf at the beautiful Southward Ho Country Club in Bay Shore, New York. The day began under a clear blue sky with an early autumn wind blowing…..which was a bit challenging to the golfers but they didn’t seem to mind at all. The day could not have been better for golfing, good times, great friends and a few laughs! The day ended with a fabulous meal, more laughs and some fantastic raffle prizes! The culmination of this wonderful day was the presentation of a check for $50,000 to CRN Board Members Marybeth Krummenacker and John Maccarone. A very special thank you goes to

The Plumbing Contractors of Long Island Association present a check for $50,000 to Marybeth Krummenacker
Hassans Ask Wedding Guests to Donate to CRN in Lieu of Gifts

By Stephanie Hassan

As of today, October 24, 2010 I’ve been married for exactly two weeks and one day. This isn’t my first marriage. My husband and I were both married before in marriages that lasted over ten years. As one would imagine, between the both of us we had respectively accumulated plenty of stuff like towels, toasters, forks and crystal bowls. As we planned our wedding we debated whether or not to bother registering anywhere, but we couldn’t do it. We couldn’t justify asking our friends and family for even more stuff that we truly didn’t need. After talking about our dilemma with a close friend, he suggested directing guests to donate to a favorite charity and the light bulb went off. My twelve year old son Austin was diagnosed with Cystinosis when he was 18 months old. I’ve always wanted to do a fundraiser for the Cystinosis Research Network but felt like I didn’t know enough people. Here was my opportunity to finally do a fundraiser for CRN! Why didn’t I think of this brilliant idea myself?

My then fiancé and I typed up our own directional cards and on the back asked guests to donate to CRN in lieu of gifts. We printed the cards off, put them in with the invitations, crossed our fingers and mailed everything off. We had hopes that maybe half our guest list would actually show up to the wedding and maybe, if we were lucky, a few of those people would donate. 127 guests showed up, and over $1,000 has been collected and counting! My husband and I could not be happier. We feel so blessed and so supported. Some of the guests that have donated are friends of my husband that I’ve never met and didn’t get a chance to see on our wedding day. Why should they care about me or my son? I can’t answer that question, but I’m thankful for these people and for this opportunity. I hope everyone gets a chance in their lifetime to experience this kind of love and support. I would encourage everyone in the Cystinosis community to find a way to get involved and help fundraise, no matter how off the wall the opportunity may seem. Since Austin was diagnosed my prayer has always been “Lord, please show us a miracle.” After the outpouring of love we’ve just experienced, I feel like he has.

Devlin Keon’s “Auntie Lil” Donates to CRN in lieu of Wedding Favors

Devlin Keon is a special little boy with cystinosis who has a very special Aunt. Elizabeth Irons, cousin to Devlin’s mom, Roisin, was married on May 29, 2010 in Winthrop, MA, and Devlin was the ringbearer. Elizabeth and Kirk Irons chose to donate $500 to CRN in lieu of wedding favors to honor Devlin. The generosity and compassion didn’t stop there. Devlin’s “Auntie Lil” as he calls her gave Devlin a pair of RayBan sunglasses for a ringbearer gift because she knows the sunlight bothers his eyes.
Sixth Annual Jack's Family Fun Day Raises $15,000 for CRN

By Christy Greeley

The Sixth Annual Jack's Family Fun Day took place on October 17, 2010. The event was a Fall Festival held at Tamarak Country School and Day Camp in Lincolnshire, IL. All proceeds benefit the Cystinosis Research Network in honor of Jack Greeley. Nearly 500 people enjoyed a beautiful Fall day and a variety of activities including music by Charizma, arts and crafts, bounce house, inflatable obstacle course compliments of the Chicago Fire, appearance by the Fire mascot "Sparky", pumpkin decorating, face and nail painting, games with prizes, hay and tractor rides, balloonist Robert Hirsch, soccer, basketball, pony rides by Patch 22, bake sale and cake walk, Big Red Fire Truck, silent auction and mystery wine bottle sale. This year's event matched last year's fundraising total of $15,000, bringing the total raised by the six Jack's Family Fun Day events to nearly $100,000. This event is run entirely by volunteers, with nearly all of the services and entertainment donated by local businesses and sponsors. We would like to particularly thank Lucia and Dave Thoensen for graciously allowing us to utilize Tamarak's beautiful grounds for the event, as well as Kathy Gordon for organizing and executing the silent auction portion of the event so well. We cannot thank our friends, family and community enough for their support!
Uncle Tim’s Fundraiser Raises $2,025 for CRN

Uncle Tim does it again! Jack Kitchen’s Uncle Tim hosted a fundraiser on October 2 and 3. Tim is the brother of Jack’s dad, Travis. Uncle Tim is from Georgia and has hosted several of “Uncle Tim’s Fundraisers” on his property. This year’s fundraiser raised $2,025 for CRN! The families participating in the Emory study were able to drive over for the day.

Kids had a blast at Uncle Tim’s Fundraiser. The kids in the Emory study were able to drive over for the day.

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!
Two Guest Bartending Events Held in Honor of Alexis and Evan LeBeau

Frank from Season 6 of “The Bachelorette” is Guest Bartender at Carlucci in Downer’s Grove, IL

Friends of the LeBeaus organized two Guest Bartending events in honor of their children, Alexis and Evan, both with cystinosis. A percentage of all sales and tips during a two hour time frame were donated to CRN at each event. Guests could make additional donations to CRN as well.

One event was August 23 in Hinsdale, IL, and it raised $2,000 for CRN.

The other event was October 23 at Carlucci Restaurant in Downer’s Grove, IL. Carlucci Restaurant arranged to have Frank from Season 6 of The Bachelorette behind the bar. The event raised $1,200 in donations and is climbing! The amount raised from tips and sales was not available at the time of publication.

Even Ghouls Support Cystinosis Research!

By Frankie McGinnis

The family of Laura McGinnis hosted a charity Haunted House (aka: SAKATS MANOR) at Drummer Boy Campground in Gettysburg, PA. Admission was free, but donations to the Cystinosis Research Network were encouraged. The haunted house was open the last 3 weekends of October. It was a collaboration of friends and family with a common interest and a common goal. The entire operation took 6 weeks to construct, decorate and prepare for the Halloween season. It would not have been possible without the contributions of the staff at Drummer Boy Campground. Many thanks to all of our friends and families for the help and love that went into making it a success. We are proud to say we raised $250.00 for CRN and educated many who attend the haunted house. We are already planning for the 2011 Haunted House.
THANK YOU TO ALL OUR DONORS!

Cystinosis Research Network would like to thank ALL of our generous donors. Without you, there would be no hope for improved treatments and ultimately a cure for cystinosis. We would not be anxiously preparing for the 2011 Family Conference where many families affected by cystinosis will meet medical experts in cystinosis for the very first time. Children with cystinosis will play together, and it will be perfectly normal to have their play interrupted to take their medicine. Without you, there would not be a talented, compassionate doctor learning to become an expert in the treatment of cystinosis. CRN could not have sponsored the 15th Congress of the International Pediatric Nephrology Association and educated doctors around the world about cystinosis. We could not seek the latest trends in advocacy at meetings such as Genetic Alliance or improve the transition to adulthood for adolescents with cystinosis. Without you, there would be no CRN.

Whether you are a large corporation or an individual donor, your contributions are important to our mission and vision. Whether you organized a sports outing or wrote a letter campaign, your fundraising efforts are vital to the continued success of our organization. **We thank you from the bottom of our hearts and wish you a happy holiday season.**

Jack Greeley’s Little League Team Wins Championship

Jack Greeley’s Grade 3-4 Cubs baseball team won the Spring Lake Sports League championship game against the Yankees on June 5 at North Park in Lincolnshire, IL. Jack’s team was 5th place after the regular season but came to life in the playoffs by scoring 51 total runs over three games in dominating, total team performances each game. Jack’s dad, Dave Greeley was one of the coaches. Go Cubs!

Participants in the Emory study take time out to pose with their nurses. Front from left to right: John Binder, Glenn Jones, Garrett Thomas, Mitchell Smith, Mason Reed, Laura McGinnis
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

Jeff and Alicia Wachter (with cystinosis), JC Wachter (without)
Minnie and Mickey (without?)
Join the Cystinosis Research Network Today!

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:
Cystinosis Research Network
302 Whytegate Ct.
Lake Forest, IL 60045

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Name________________________________________________________________________
Street________________________________________________________________________
City & State_______________________________________ Zip Code__________ Country ____
Phone__________________________Fax_______________________Email________________
Name of Child / Adult / Acquaintance / Patient affected with cystinosis:________________________

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.
Please Support CRN’s Mission with Your Donation

☐ 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 Good-Search 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 Good-Shop 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!
United Way Contribution Guidelines

Identify the Cystinosis Research Network, Inc. as the agency you want to receive your contribution through the United Way Donor Choice Program.

<table>
<thead>
<tr>
<th>Agency Name</th>
<th>The Cystinosis Research Network, Inc.</th>
</tr>
</thead>
<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.