Sigma-Tau Pharmaceuticals, Inc. Receives FDA Approval of CYSTARAN™

GAITHERSBURG, MD, October 4, 2012—Sigma-Tau Pharmaceuticals, Inc. announced today that the Company has received approval from the U.S. Food & Drug Administration (“FDA”) for CYSTARAN™ (cysteamine ophthalmic solution) 0.44%, a topical ophthalmic therapeutic, developed in partnership with the National Institutes of Health (“NIH”), for the treatment of patients suffering from corneal cystine crystal accumulation as a result of cystinosis. CYSTARAN™ is designated an Orphan Drug in the U.S. and has been granted seven years of market exclusivity.

Cysteamine is a cysteine-depleting agent which lowers the cysteine content of cells in patients with cystinosis. However, when orally administered, cysteamine does not reach the cornea and is therefore ineffective in reducing the ocular effects of cystinosis. CYSTARAN™ is for topical ophthalmic use and is indicated for the treatment of corneal cystine crystal accumulation in patients with cystinosis. As a result, CYSTARAN™ now represents the only FDA-approved ophthalmic treatment for this condition.

"As a Company dedicated to the development and commercialization of novel therapies that address the unmet medical needs of a wide range of rare disease patients, we are delighted to announce the approval of CYSTARAN™," noted Dave Lemus, Chief Operating (continued on page 4)

CRN Family Conference: Heroes Among Us

July 18-20, 2013 in Washington, D.C.

The Cystinosis Research Network is pleased to announce the 2013 Family Conference "Heroes Among Us" will be held July 18-20, 2013 at the Marriott Wardman Park Resort in Washington, D.C.

Please plan to join us to reconnect and learn more about new research findings, meet and renew friendships with other families, have the opportunity to participate in research studies, and interact one on one with many of the world's expert clinicians treating and researching cystinosis today. (continued on page 19)
Message from the President

Jeff Larimore

“A Time for Introspection”

One of the many acknowledged strengths of the Cystinosis Research Network is its recognition that transition within its Board membership is a time of evaluation and rejuvenation. With great pride and commitment, I am pleased to have been nominated and accepted by CRN’s Board to become the organization’s next President.

Board membership transition provides a unique opportunity in the life of an organization for it to step back and reflect, not just on the transition itself, but more broadly on mission and goals. By preparing itself to welcome and orient to new voices and personalities, the organization will necessarily review its finances, policies, procedures, and operations.

For the past five years, the Cystinosis Research Network has been valiantly led by Christy Greeley. The number of accomplishments that CRN has achieved while Christy was President cannot be fully documented in a short summary. Christy, our gratitude and appreciation for the energy and commitment that you and the Greeley family have expended likely will never be fully expressed without missing a detail or two. You have set the bar high and I am excited to have received the baton to lead the Cystinosis Research Network. We are grateful that you will be continuing as Executive Director.

Some personal background on the Larimore family is that my wife Katie and I have been married for 20 years and have four children, Jessica (17), Caroline (16), Daniel (11), and Sarah (7). Sarah was diagnosed with cystinosis in 2006. Cystinosis affects everyone in this household and Katie and I cannot express how much assistance and love all of our children exert in its management.

When the Nominating Committee inquired if I would have an interest in becoming CRN’s President, I had to assure myself that I was the right person for the position. I’ve been part of the organization over the last 18 months as Treasurer but knew that the new responsibilities would be broader and more diverse. The support extended by the members of the incoming Executive Committee calmed my anxieties fairly quickly.

The previous editions of The Cystinosis Advocate have also proven to be an invaluable resource for my transition. A warm-hearted thank you to Paula Shal in making the newsletter the “printed voice” of the Cystinosis Research Network. We are excited about the editorship transition to Jessica Jondle.

In reviewing Christy’s previous messages, I noted a series of remarks that highlight what the Cystinosis Research Network pursues to accomplish. At this time for introspection, the Board of Directors strives to have:

- new families diagnosed earlier
- better treatments for our children
- adults living long and productive lives
- promising and innovative research occurring worldwide
patients diagnosed and treated appropriately around the globe
clinical specialists committed to a career spent caring for and researching cystinosis
more families advocating for their children and adults advocating for themselves
CRN continuing to engage, advocate, and represent global rare disease communities with spirit and integrity

These eight fundamentals are byproducts of executing the vision and mission statement of the Cystinosis Research Network. The new Board recognizes that our role in facilitating collaboration within the cystinosis community will continue to be one of our most profound 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:

Research—Colleen Hammond has returned to become the V.P.—Research. Colleen is a co-founder of the Cystinosis Research Network and a past President. Recent news in Research includes Sigma-Tau receiving FDA approval of Cystaran, the liquid cysteamine medication used to dissolve cystine crystals in a patient’s corneas, along with dynamic progress in research grant programs. Colleen is also the Chairperson for the 2013 Family Conference to be held July 18-20, 2013 in Washington, D.C. Research updates will be provided throughout the newsletter.

Development—Tim Miller, the first Board member of CRN not to have a family member affected by cystinosis, has agreed to become V.P.—Development. Tim is actively communicating with families and sponsors to enhance member involvement and fundraising opportunities. Development updates and calls to action will be highlighted throughout the newsletter.

Family Support—Pam Woodward will continue to anchor the role of V.P.—Family Support. Family Support is the first point of contact for new families looking for information on how to manage through the demands of daily life with cystinosis. Pam is also active in the preparation details of the 2013 Family Conference.

Education & Awareness—Jessica Jondle has agreed to carry the torch as V.P.—Education & Awareness. This Board position has multi-dimensional responsibilities including editorship of the bi-annual newsletter, participation and exhibition at medical conferences, and communication with families who have questions relating to cystinosis. Jessica recently released a memoir called *Roller Skating with Rickets and Other Paradoxes of Life with Genetic Disease*. The Foreword makes a profound statement: “If you don’t believe in miracles, you should.” Education & Awareness updates will be highlighted throughout the newsletter.

Financial—Jenni Sexstone has become the new Treasurer for CRN. Jenni will coordinate the preparation of CRN’s financial statements, operating budget and forecasts, and communicate directly with outside accountants, auditors, and tax preparers. The 2011 audit and tax return, Form 990, have been completed and will be posted on CRN’s website for viewing in November. A financial update is presented in this newsletter.

Secretary—Terri Schleuder has become the new Secretary for CRN. Terri will assist each committee with service support and will be the official record keeper for CRN.

Each Board Member and Committee Member for the Cystinosis Research Network feels deeply in their hearts that their involvement is for a reason and has purpose. CRN always needs to have more families involved. We have only scratched the surface of its potential and CRN’s accomplishments have made a difference in the United States and internationally. This organization and its dedicated people have meant the world to the Larimore family.

Warmest Regards,
Jeff Larimore
President

Jeff & Katie Larimore
FDA Approval of Cystaran™ Eye Drops
(continued from page 1)

Officer of Sigma-Tau. “This new medicine will offer physicians the only FDA-approved treatment for patients with corneal cystine crystal accumulation, many of whom are children and whose lives are seriously impacted by this debilitating chronic condition.”

“This is an important advance for children and adults who suffer from cystinosis,” stated William A. Gahl, M.D., Ph.D., Clinical Director, National Human Genome Research Institute, NIH. “FDA approval of this drug represents the culmination of a longstanding collaboration among the National Eye Institute, the Eunice Kennedy Shriver National Institute of Child Health and Human Development, the National Human Genome Research Institute and Sigma-Tau Pharmaceuticals. It also has involved invaluable cooperation from cystinosis advocacy groups—the Cystinosis Research Network, the Cystinosis Foundation and the Cystinosis Research Foundation.”

The CYSTARAN™ project was supported in part by an Orphan Drug grant from the FDA.

The clinical safety and efficacy of CYSTARAN™ was previously evaluated in controlled clinical trials conducted by the NIH, in approximately 300 patients. Results of these studies support the use of ophthalmic cysteamine as an effective treatment of corneal cystine crystals. The most frequently reported ocular adverse reactions, occurring in ≥ 10% of patients, were sensitivity to light, redness, eye pain/irritation, headache and visual field defects.

CYSTARAN™ is planned to be available in the future through specialty pharmacy distribution channels.

Cystaran™ Frequently Asked Questions

What is Cystaran™ (cysteamine ophthalmic solution) 0.44% and what is it indicated for?
Cystaran™ (cysteamine ophthalmic solution) 0.44% is an ophthalmic solution indicated for the treatment of corneal cystine crystal accumulation in patients with cystinosis.

When will the product be available?
Sigma-Tau Pharmaceuticals plans to have Cystaran™ available in the US in 2013. Sigma-Tau is currently working on a program to make the product available to patients outside of the U.S.

When was Cystaran™ approved?
October 2, 2012.

How much will Cystaran™ cost and will it be reimbursed?
At this time pricing is not available. Pricing for Cystaran™ will be available at the time of product launch.
We expect Cystaran™ to be covered by most medical and prescription drug plans in the US. Our goal is to ensure that patients who need Cystaran™ are able to obtain the medicine. We will have assistance programs in place when product is available.

Can I still use the compounding pharmacy?
You should discuss this with your prescribing physician.

What should I do with the drops I already have?
Continue to use the drops as directed by your physician.

Who do I go to to get a prescription for Cystaran™?
Please speak with your treating physician.

The studies used 0.55% cysteamine hydrochloride eye drops. Why is Cystaran™ 0.44%?
Cystaran solution contains 6.5 mg/mL of cysteamine hydrochloride which is equivalent to 4.4 mg/mL of cysteamine (0.44%). The FDA required us to label Cystaran by its cysteamine content, thus the label refers to the 0.44% cysteamine.

Can I still get my eye drops from NIH?
Yes, NIH plans to continue to supply cysteamine eye drops until Cystaran™ becomes commercially available. Please contact the research coordinator, Alana Temple at (301) 402-1369, for additional information about obtaining eye drops through the NIH.

Important Safety Information for Cystaran™
The most frequently reported ocular adverse reactions, occurring in ≥ 10% of patients, were sensitivity to light, redness, eye pain/irritation, headache and visual field defects.

Please see the prescribing Information available at www.cystaran.com.
For additional questions, please contact Lesli King, Senior Manager of Patient Affairs, at (301) 670-5450 or email Lesli.King@sigmatau.com.

NIH Growth Hormone Study for Adults with Cystinosis Experiencing Muscle Weakness
CRN is working to advocate and support research for adults with cystinosis. We are collaborating with the NIH to conduct a growth hormone study for adults with cystinosis who are experiencing muscle weakness. Progress regarding the initiation of the study continues. The study has been approved by the NIH scientific review approved and is awaiting IRB and safety approval. We will keep you updated regarding the start of the study.
The Cystinosis Research Network was honored to have been selected as the family advocacy co-sponsor and attend the combined 4th Cystinosis Annual Meeting and the 6th Cystinosis Symposium of the Mexican Association of Cystinosis, held August 16-17, 2012 in Mexico City, Mexico. Victor Gomez, President of the Asociación Mexicana de Cistinosis, and Dr. Leticia Belmont, supporting medical director, narrated the well-attended symposium. This event brought together cystinosis patients and their families, plus medical students, doctors, and researchers from Mexico, Europe, and the United States.

Representing CRN were Board members Jeff Larimore and Elva Smith, and professional co-sponsors were Alexion Pharmaceuticals, Sigma Tau, and Raptor Pharmaceuticals. The Asociación Mexicana de Cistinosis certainly achieved its goal of providing up-to-date information on improvements in cystinosis treatment, current research, and the raising of awareness of cystinosis in Mexico.

The first day program, which was the 4th Annual Cystinosis Meeting, focused primarily on the cystinosis patient and patient issues, including medical treatment and daily management of life with cystinosis. Among those giving presentations were Drs. Leticia Belmont, Ewa Elenberg, Elena Levtchenko, Craig Langman, and Vanessa Bosch. Additional presentations were given by Sara Guillen Nut, Iliana Espinoza, Diana Trovar, and Jeff Larimore. Victor Gomez gave an update on the progress and goals of the organization. Some of the topics covered were the daily challenges of life for the cystinosis patient, side effects of the drug cysteamine, kidney transplantation, and eye and dental care.

Following the program presentations, the Mexican cystinosis patients were given medical check-ups and evaluations by the attending medical committee. A stimulating question-and-answer session, which touched on many topics, was conducted by the cystinosis families. Jeff Larimore and Elva Smith participated in the Q&A session and provided some insight into how to improve communication, organizational development, and fundraising.

Friday’s session, which was the 6th Cystinosis Symposium, was held in the prestigious Mexican Academy of Surgery, Congress Unit, National Medical Center SXXI auditorium and brought together a most impressive representation of the medical community in Mexico. Presentations were directed...
Cystinosis Mexico Symposium (cont.)

towards research, diagnosis, and general medical issues on cystinosis. In addition to the speakers from the previous day, papers were given by Drs. Eduardo de la Teja, Juan Carlos Jaurez, and Miguel Angel Alcantara as well as Drs. Elenberg, Levchenko, and Langman. Medical professionals in attendance were from the fields of nephrology, gastroenterology, genetics, ophthalmology, dentistry, and nutrition.

Cystinosis Mexico has made, and is continuing to make, significant strides in promoting awareness of cystinosis throughout Mexico. The group has worked hard to overcome the challenges of receiving Cystagon and it awaits the availability of cysteamine eye drops. In addition to being the strong voice of the cystinosis community, the organization provides much-needed family support and is instrumental in educating the Mexican people, medical community, and elected officials of the difficulties of living with and caretaking for patients with cystinosis.

If you would like additional information regarding Cystinosis Mexico, visit their website at cystinosismexico.com, contact Victor Gomez at victor.gomez@cystinosismexico.com or to request general information, submit an email to info@cystinosismexico.com.

Update from Asociación Mexicana de Cistinosis

by Victor Gomez—President, AMC

The 4th Annual Cystinosis Meeting and 6th Cystinosis Symposium in Mexico City represented the very first time that both events have been held together. We were delighted to have special guests from the Cystinosis Research Network, Jeff Larimore and Elva Smith, as well as doctors from nearby cities, patients, and their families attending these meetings.

The 4th Annual Meeting was focused on family-patient support, information, and motivation, with topics such as success in life, cysteamine effects, and the value of kidney transplant. At the end of this day, speaker Jeff Larimore talked about Power in Patient Advocacy Organizations.

The 6th Cystinosis Symposium was intended just for physicians. Some of the focus was on the natural history of cystinosis, molecular biology, and new treatments. Finally, cystinosis experiences in Europe and Mexico and research were shared by Dr. Elena Levchenko, Dr. Leticia Belmont, and Elva Smith.

We appreciate Cystinosis Research Network, Sigma Tau, and Raptor Pharmaceutical for sponsoring cystinosis meetings in Mexico City.
Financial Update

by Jenni Sexstone—Treasurer

We are pleased to present the financial results of the Cystinosis Research Network through September 30, 2012. These financials represent the great efforts of all CRN members: patients, parents, family, friends, and researchers focused on educating the public and medical communities and improving quality of life of those with cystinosis.

For the nine months ending September 30, 2012, total income secured of $268,000 and was approximately 11% less than the same period in 2011. The decline is primarily driven by timing of grants received from Sigma-Tau Pharmaceuticals and a 5% decline in overall fundraising. Total donations received from public organizations such as United Way, Network for Good, and Facebook were approximately $42,000, which is 13% above 2011.

Administration, registration, and travel costs associated with Education & Awareness amounted to approximately $11,000 through September 2012. Expenses for the 2011 CRN Family Conference are driving the large decline in Education and Conference spending over the same period for 2011. General operating expenses for CRN through September 2012 of $49,000 were approximately $7,000 less as compared to the nine months ended September 2011.

A primary objective of the Cystinosis Research Network is to provide funding for research grants. These programs focus on identifying the underlying effects of cystinosis and expanding outreach and education domestically and internationally. Total grant payments of approximately $272,000 have been made through September 2012. The institutions that have received support from CRN in 2012 include Baylor University; University of Michigan; Cairo University, Cairo, Egypt; University Hospitals, Leuven, Belgium (joint funding with Cystinosis Ireland); University of Sunderland, Sunderland, Scotland; and VU University Medical Center, Leuven, Belgium.

For the period ending September 30, 2011, approximately $319,000 of grant commitments were paid. Throughout the newsletter will be more detailed information regarding the status and findings of each of the research programs in progress.

Among the challenges ahead for the Cystinosis Research Network is the funding of the research grant commitments that have not yet been progress billed to the organization. For the remainder of 2012 and through 2013, $188,000 of research grant commitments could be paid out. We are also heavily into the planning stages of the 2013 Family Conference, which also requires significant financial resources. In order for CRN to remain focused on providing continued education/awareness of and improving quality of life of those with Cystinosis through funding of ground-breaking research, it is imperative for all CRN members to ensure sustainability by remaining focused on continued and new fundraising and donation efforts.

A comparable financial recap for Cystinosis Research Network for the nine months ended September 30, 2012 and 2011 is included on the following page.
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<thead>
<tr>
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<th>Jan-Sep 12</th>
<th>Jan-Sep 11</th>
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<tr>
<td><strong>Total Income</strong></td>
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<td><strong>Total Expense</strong></td>
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<td><strong>Net Operating Income/(Loss)</strong></td>
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<td><strong>Cash on Hand—Jan 1</strong></td>
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<td><strong>Net Change in Cash</strong></td>
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<td><strong>Cash on Hand—Sep 30</strong></td>
<td>$103,941</td>
<td>$173,324</td>
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</table>

Accenture Contributes $10,000 in support of CRN’s Vision and Mission!

Accenture is a global management consulting, technology services, and outsourcing company that combines unparalleled experience, comprehensive capabilities across all industries and business functions, and extensive research on the world’s most result-oriented companies. Accenture collaborates with its clients to help them become high-performance businesses and governments. Accenture has approximately 249,000 professionals serving clients in more than 120 countries.

Accenture has six core values:
- Client Value Creation
- Best People
- One Global Network
- Integrity
- Respect for the Individual
- Stewardship

It is under the Stewardship value where the aspiration of being an exemplary corporate citizen and encouraging the active involvement of employees in the communities where Accenture’s professionals work and live that provided the basis for the contribution. In conjunction with Accenture’s Hispanic Executive Summit, Accenture has committed to make a charitable contribution to an eligible non-profit organization on an annual basis. This year, Accenture selected the Cystinosis Research Network to receive its generous donation of $10,000. Christy Greeley, CRN’s Executive Director, provided an overview of CRN’s activities, mission, and vision and accepted the contribution on behalf of the organization. Christy expressed her deepest gratitude to Accenture for their support of CRN’s programs, and was extended an invitation to participate in the evening’s bowling tournament. Christy’s team was in contention throughout the evening and eventually placed third.
Family Support Update

by Pam Woodward—Vice President, Family Support

The Family Support Committee, along with the conference subcommittees, are busy at work preparing for the upcoming CRN Family Conference to be held in Washington, D.C. on July 18-20, 2013. The agenda is filled with presentations covering the various topics related to cystinosis—everything from the history of cystinosis to past favorites such as the medical and patient panels and of course the popular breakout sessions with more specialized topics. Looking forward to seeing everyone in Washington, D.C. in July!

The Family Support and Research Committees have also just finished the Cystinosis Parent Handbook; it is in the final editing stages now and will be available soon. We are excited to be publishing this guide and believe it will be very useful to the members of the cystinosis community as they deal with the everyday trials of living with cystinosis.

The CRN Family Support Committee supported our community by sponsoring a dinner at the Midwest regional family gathering which was held near Indianapolis, Indiana on July 20-22, 2012. The family gathering was hosted by the Gard family. (See the article from Dena Gard, next page).

Hope everyone has a wonderful holiday season and a happy 2013.

Pam Woodward (pwoodward@cystinosis.org)

Feature Family: The Sausman/Kelly Family

June 9, 2012

Little James Carson Sausman was born 5lb 3oz on April 29, 2009, six weeks early but very healthy, even coming home in the normal 24-hour period that most infants do. He came home to a very full house. He has five siblings who now range in age from 10-19. Once home, James did very well for the first 9-10 months. He was growing and seemed healthy. Then suddenly he quit growing and an array of other symptoms began, such as loss of appetite, water guzzling, and constipation. We have a very good general pediatrician who always knew that there was something wrong; we just had to find out what it was. After eight months he sent us to WVU to see a nephrologist, who after a week-long hospital stay and many tests diagnosed James as having cystinosis. The doctor immediately started him on Cystagon, along with many other medications.

It has been a year ago this week since we first heard the word cystinosis and I’m happy and elated to say James is a thriving, happy-go-lucky three year old and is doing much, much better. He loves being outside with us playing in our garden, riding his Big Wheel, petting his kitty, and being a part of everything he can.

Sincerely yours,

The Sausman/Kelly family
Jim, Kate, James, Jonas, Emily, Sarah, Lindsey, and Ashley
Annual Midwest Cystinosis Family Gathering

by Dena Gard

The annual Midwest Cystinosis Family Gathering was held in Plainfield, Indiana on July 20-22, 2012 at the Wingate Hotel. The fun started off on Friday evening as families arrived with a pizza supper sponsored by the Cystinosis Research Network. The fun continued through the weekend as an informal meeting was held on Saturday morning featuring our guest speakers, Lawrence and Kristy from Raptor Pharmaceuticals. They explained the new drug RP103 (time-release or 12-hour cysteamine) and patient support program they are starting called RaptorCares. The families were allowed to ask questions and the Raptor representatives were very generous in giving us answers. Lunch was shared by the group as we went to the Metropolis Mall in Plainfield and ate at Teddy’s Burger Joint. After lunch some went shopping and some went to an amusement park, where they enjoyed go-karts and mini golf. That evening Raptor sponsored our evening meal at the Stone Creek Dining Company. After dinner some went to watch the new Batman movie while others went back to the hotel to visit in the meeting room, which was sponsored by the Cystinosis Foundation. Sunday morning we said our goodbyes and a couple of the families went to the Indianapolis Zoo before parting ways. Seven families were represented from several states, including Iowa, Minnesota, Missouri, Illinois, and Indiana. We would all like to thank the Cystinosis Research Network, Cystinosis Foundation, and Raptor for helping to make our weekend a great success!

AST Announces Newly Elected President

Congratulations CRN Scientific Review Board member Dr. Roslyn Mannon on your election as President of the American Society of Transplantation! Your dedication to research, the education of others in the field, and ongoing participation on review boards such as ours is truly inspiring. We think this was an election well deserved. Thank you for all of your hard work!

For more information on the AST and Dr. Roz Mannon, please visit http://www.a-s-t.org/news/ast-welcomes-new-board-members.
Research Update

by Colleen Hammond—Vice President, Research

A major focus of the Cystinosis Research Network continues to be a determined effort to secure a promising future for the cystinosis community through the support and funding of research grants that lead to improved treatments and ultimately a cure for cystinosis. CRN’s Scientific Review Board, comprised of the leading cystinosis experts, guides CRN in funding the research studies. CRN has funded nearly $3 million total in research grants and fellowships. Information regarding these research grants, progress reports, funding and final reports is provided to the cystinosis community via our web site www.cystinosis.org.

Current Research Studies:

Neveen Soliman, MD and Elena Levchenko, MD, PhD, Center of Pediatric Nephrology and Transplantation, Cairo University, Egypt and Department of Pediatric Nephrology University Hospitals, Leuven, Belgium, “Genotype-Phenotype in Egyptian Patients with Nephropathic Cystinosis.” Total award: $15,750.

Martine Besouw, MD, Department of Women and Child Laboratory of Pediatrics, University Hospitals, Leuven Belgium, “Study of skin changes in cystinosis patients under cysteamine therapy”, Mentor: Elena Levchenko, MD, PhD. Total award: $99,400.00. This grant is a joint funding collaboration between Cystinosis Research Network and Cystinosis Foundation Ireland.

Maya Doyle, LCSW, Children's Hospital at Montefiore, Division of Pediatric Nephrology, New York University, "Cystinosis in Emerging Adulthood." Total grant award: $17,200.

Jess G. Thoene, MD, Director, Biochemical Genetics Laboratory, Active Professor Emeritus of Pediatrics, University of Michigan, "Continuation of Feasibility of Cystinosin Replacement Therapy in Cystinosis.” Total award: $202,500.00.

Rosaleen Anderson, PhD, Sunderland Pharmacy School, University of Sunderland, "Proteomic investigation of cystinotic cells and the effects of cysteamine treatment." Total award: GPB 80,926.00 or $130,366.00.

Henk J. Blom, MD, M.M.C. Wamelink, and E. Levchenko, MD, VU University Medical Center, Amsterdam, The Netherlands: “Newborn Screening of Cystinosis.” Total award: 45,000 euros (approximately $67,500.00).

Francisco Emma, MD and Anna Taranta, PhD, Bambino Gesu Children’s Hospital and Research Institute, Rome, Italy: “Functional Characterization of Cystinosin-LKG.” Total grant award: $114,480.00.

Ewa Elenberg, MD, Texas Children's Hospital, Houston, TX "Quality of Life in Cystinosis Patients." Total award: $21,000.00.
New UCSD Online, Questionnaire-Based Study for Adults with Cystinosis

The Pediatric Neurology Research Group at the University of California, San Diego (UCSD) is conducting the following research study: Outcome and Quality of Life in Adults with Cystinosis

About the Study: Angela Ballantyne, Ph.D. and Doris Trauner, M.D., of the UCSD School of Medicine, are conducting a questionnaire-based research study to learn more about outcome and health-related quality of life in adults with the genetic-metabolic disorder, cystinosis. The purpose of this study is to gather vital information on outcome and quality of life in the ever-expanding population of adults living with cystinosis. The information obtained from this study can also be used to weigh the risks versus benefits of new therapies or treatments for cystinosis.

What the Study Involves: This is an online, questionnaire-based study that examines quality of life and outcome in adults with cystinosis. Participants will be asked to log in to a secure, online assessment center and complete a series of questionnaires that will take approximately 1 to 2 1/2 hours. Questions will be asked regarding demographic background, health, medical issues, quality of life, and outcome in cystinosis. Participation in the study is strictly confidential. Data from all participants will be combined, and you will not be identified by name in any publication or teaching activities.

Who May Participate: Two groups of participants are being recruited to take part in this online study. You are eligible to participate if:
(a) you are an adult over 18 years of age living with cystinosis, OR
(b) you are a parent or nearest living relative of an adult who had cystinosis and passed away within the past 10 years.

Participation in this research study is completely voluntary. If you would like to participate in the Cystinosis Adult Outcome study or if you would like more information, please contact 858-822-6800 or email Jennifer Crowhurst at jcrowhurst@ucsd.edu.

New Clinical Research Study at UCLA Medical Center
Now Enrolling Patients

New Clinical Research Study at Ronald Reagan UCLA Medical Center

The first allogeneic (donor) stem cell transplant was approved by the UCLA Investigational Review Board (IRB) and the recruitment process has begun. The stem cell protocol was designed by a team of leading stem cell experts and cystinosis experts who formed the CRF Cystinosis Gene Therapy Consortium. The Consortium members are: Dr. Corinne Antignac (Hospital Necker, Paris, France), Dr. Stephanie Cherqui (Scripps Research Institute), Dr. William Gahl (NIH), Dr. Don Kohn (UCLA), Dr. Ted Moore (UCLA), Dr. Dan Salomon (Scripps Research Institute), and Dr. Jerry Schneider (UCSD).

If you would like additional information about the pilot study, please view the ad on the CRF web site at http://www.natalieswish.org/files/11-003240CystinosisUCLASStudy-5-8-12.pdf, or visit the Cure Cystinosis International Registry, CCIR. If you are interested in learning more, please contact your primary care physician and ask him/her to contact Zoe Solsby at CRF. Zoe's email address is zsolsby@cystinosisresearch.org and her phone number is (949) 223-7610.
The Cystinosis Research Network was honored to participate in The Celebration of Science, September 7-9, 2012 in Washington, D.C., an intensive three days focused on jumpstarting a new era of scientific discovery where the economic and social benefits of scientific research and how it improves lives around the world were highlighted. [http://celebrationofscience.org/](http://celebrationofscience.org/).

The meeting was sponsored by the Milken Institute, a non-partisan think tank chaired by Michael Milken whose mission is to improve lives around the world by advancing innovative economic and policy solutions that create jobs, widen access to capital, and enhance health. FasterCures, an organization run by the Milken Institute, works alongside patient advocates, researchers, investors, policymakers, and other leaders in medical research and development to stimulate innovative collaborations across all sectors—academia, government, industry, investors, and nonprofits.

The weekend included panels and brief presentations, a full day of activities on the campus of the National Institutes of Health and a FasterCures working meeting on accelerating innovation in the biosciences, as well as a special evening at the Kennedy Center to honor several luminaries who have made remarkable contributions and support of advancements in science:

**Medical & Research Luminaries:**
- S. Ward Casscells, Tyson Distinguished Professor of Medicine and Public Health, University of Texas at Houston
- Francis Collins, Director, National Institutes of Health
- Story Landis, Director, National Institute of Neurological Disorders and Stroke
- Christopher Logothetis, Chair, Department of Genitourinary Medical Oncology, MD Anderson Cancer Center
- Jonathan Simons, President and CEO, Prostate Cancer Foundation
- Rudolph Tanzi, Joseph P. and Rose F. Kennedy Professor of Neurology, Harvard Medical School
- Harold Varmus, Nobel Laureate; Director, National Cancer Institute
- James Watson, Nobel Laureate; Chancellor Emeritus, Cold Spring Harbor Laboratory
- Elias Zerhouni, President, Global R&D, Sanofi; former Director, National Institutes of Health
Corporate Luminaries:
• Margaret Anderson, Executive Director, FasterCures
• Michael Milken, Chairman, Milken Institute; Founder, Faster Cures
• Freda Lewis-Hall, Executive Vice President, Chief Medical Officer, Pfizer Inc.

Patient Advocacy Luminaries:
• Robert Beall, President & CEO, Cystic Fibrosis Foundation
• Nancy Brinker, Founder and CEO, Susan G. Komen for the Cure, former U.S. Ambassador to Hungary
• Deborah Brooks, Co-founder & Executive Vice Chair, The Michael J. Fox Foundation for Parkinson’s Research

Government Luminaries:
• Eric Cantor, US Representative, Virginia (Majority Leader)
• Daniel Inouye, US Senator, Hawaii (Chairman, Committee on Appropriations)
• Nancy Pelosi, US Representative, California (Minority Leader)
• Connie Mack III, Former US Senator, Florida
• Harry Reid, US Senator, Nevada (Majority Leader)

Participants included approximately 1,000 current and former elected officials, heads of government agencies, major philanthropists, leaders of academic research centers, patient advocacy groups, distinguished scientists, and industry CEOs. Key objectives of networking, learning, and the advancement of the agenda for cystinosis and rare disease by meeting with researchers from a variety of agencies at the NIH, medical schools and universities, private research organizations such as the Cold Spring Harbor Laboratory, other patient advocacy group leaders, and Board members of FasterCures was achieved.

The commitment of the Milken Institute and FasterCures to forging bipartisan consensus about the importance of bioscience in general and the NIH in particular is quite extraordinary and we continue to seek opportunities to participate and support these efforts.

Keith and Billy Croce Memorial Scholarship

The Keith and Billy Croce Memorial Scholarship has been established with the Cystinosis Research Network (CRN) to provide financial assistance to families affected by cystinosis. The funds will help with travel expenses for those who wish to attend CRN Family Conferences. The conferences are held at different locations every two years and draw families worldwide. The event allows families to share knowledge, offer support to one another, learn more about new research findings, receive updates from cystinosis organizations from around the world, meet and renew friendships with other families, have the opportunity to participate in research studies, and interact one-on-one with many of the world’s expert clinicians treating and researching cystinosis.

Donations can be sent to:
Cystinosis Research Network
302 Whytegate Court
Lake Forest, IL 60045 USA

Or made through PayPal by visiting:

Checks payable to CRN—Keith and Billy Croce Memorial
CRN Participates in U.S. Conference on Rare Diseases and Orphan Products


Christy Greeley and José Morales attended the U.S. conference on Rare Diseases and Orphan Products on behalf of CRN as invited panelists. The event was sponsored by NORD, DIA, FDA, NIH, Eurodis, and the Duke Department of Pediatrics. This is an annual meeting where all stakeholders—patients, patient organizations, researchers, drug and device companies, investors, thought leaders, and government—meet to focus on rare diseases and orphan products research, development, and access.

CRN’s specific objectives in participating in this year’s event included increasing insights and awareness of current rare disease issues/developments; broadening visibility and awareness with major relevant pharmaceutical companies and rare disease organizations of CRN and cystinosis; strengthening relationships with our partners NORD, Orphan Europe, Sigma Tau, and other at-large advocacy groups; and advancing market awareness via press and public education publications channels.

Christy Greeley participated on the patient advocate panel to kick off the conference, which was attended by several members of the health care press. Christy was one of six patient group representatives who spoke to the struggles and successes in navigating the drug development process. Christy’s remarks focused on the long journey and eventual success of the approval of the cysteamine eye drops. She was the last speaker and was introduced as a "rare disease success story." An article ran in the Rare Disease report online highlighting the panel.

José Morales was one of two panelists discussing the "Internationalization of the Rare Disease Community." Moderator was Steve Usdin, Washington editor, BioCentury and co-host, BioCentury This Week. Paul Perreault, President CSL Behring, spoke on the industry perspective on the topic, while José addressed the emerging importance and enabling contributions of advocacy groups to internationalization. The session was very well received and provided expanded visibility for CRN and cystinosis.

Both Christy and José have been interviewed for upcoming articles related to the topics presented at this meeting. This conference is one of the many important advocacy events CRN attends in order to represent the cystinosis community and ensure that our viewpoints and voice are heard by the rare disease community.
CRN Cystinosis Awareness Bracelets Are Here!

by Terri Schleuder

The idea was born almost a year ago while eating breakfast with a dear friend at our favorite restaurant. We’d been discussing fundraising ideas for CRN when she mentioned her recent endeavor of making “Bracelets for a Cause.” She had my complete attention. For the last ten years or so she’s made beautiful, unique handmade jewelry. I have several of her pieces including two sets designed for me for my oldest sons’ weddings.

I asked her if she’d consider designing a Cystinosis awareness bracelet for CRN. She excitedly agreed. She gifted me with one in April. It is beautifully made with sparkling Swarovski crystals and silver plated beads and accents. Each bracelet is 7 inches long. ( Longer ones can be custom ordered.) We negotiated a price that would allow a $10.00 per bracelet donation to CRN and still be affordable.

Currently there are a limited number of CRN Cystinosis Awareness Bracelets available for purchase at $20.00 each. More will be made as orders dictate. A donation of $10.00 for each bracelet sold will go to CRN to support its mission and vision.

There are two ways to obtain your bracelet. You can fill out the order form at the bottom of this article and mail it to me with a check for $20.00 made out to Cystinosis Research Network. Upon receiving the check and order form, I will mail you the bracelet. Or you may e-mail your request and contact information to: tschleuder@cystinosis.org. Again when I receive the check to CRN for $20.00 per bracelet I will mail you the bracelet. If you order by email please put CRN Bracelet in the subject line.

Remember, initially there is a limited supply, so don’t wait to order and receive your beautiful, handcrafted CRN Cystinosis Awareness Bracelet. Order now!

CRN Cystinosis Awareness Bracelet Order Form

Yes, I would like to order ______ CRN Cystinosis Awareness Bracelets for $20.00 each.

Enclosed is a check to Cystinosis Research Network for: $ ______

Name: ____________________________________________________________

Address: __________________________________________________________

______________________________________________________________

E-mail: ____________________________ Phone: ________________

Mail to:

CRN Bracelets
c/o Terri Schleuder
40472 Franklin Mill
Novi, MI 48375
Raptor: Collaborating on a Patient Support Program

With the support of the Cystinosis Research Network (CRN), Raptor Therapeutics (Raptor) has been in touch with many members of the CRN community to learn how the company can support those living with cystinosis. Many of you had a chance to provide your input. Raptor is excited to announce a patient support program for families living with cystinosis, called RaptorCares™. Below are some frequently asked questions.

Q: Has Raptor filed with the Food and Drug Administration (FDA)?
A: Raptor submitted its marketing application to the FDA on March 30, 2012. The FDA has 10 months to review the application, and Raptor is expected to get a response by January 30, 2013.

Q: What is RaptorCares?
A: RaptorCares is a patient support program created by Raptor. The program, which was created with input from the cystinosis community, provides tools, support, and information to help patients and caregivers better manage cystinosis.

Q: How will RaptorCares assist families?
A: RaptorCares aims to provide tools, information, and support to families living with cystinosis.

Tools and information that are currently available to RaptorCares enrollees and other content to be available soon include:

- Cystinosis management brochure: a helpful tool for those new to cystinosis or all who need a refresher, in one easy-to-read booklet, all in one space
- Emergency room protocol and information sheet that can be shared with doctors and nurses in the emergency room
- Resources to help manage cystinosis at all ages, including:
  - Information about cystinosis that can be shared with others (such as schools and childcare providers)
  - Email series for parents who are learning to manage cystinosis
  - Cystinosis management materials for adults and adolescents

Additionally, if Raptor’s product is approved by the FDA, RaptorCares will eventually help facilitate the reimbursement process for U.S. patients.

Q: What can RaptorCares offer today?
Enroll in RaptorCares today and you will receive a cystinosis guide for schools, the emergency room protocol, and other resources included above. You will also receive a free cooler for use away from home and a copy of the 2011 global cystinosis survey, which notes the triumphs and challenges of those living with cystinosis around the world. Most importantly—you’ll get access to the most up-to-date information from Raptor!

Q: Why is Raptor motivated to create RaptorCares?
A: Raptor has created RaptorCares because the company has made a commitment of education and support, beyond potentially providing a treatment option.
Q: How can people enroll for RaptorCares?
A: People can enroll for RaptorCares by visiting www.RaptorCares.com or by calling 1.855.888.4004. Fully enrolling in RaptorCares will require a follow-up call, but the process will take less than 10 minutes and all information provided will remain confidential. Patient privacy is very important to Raptor. The full privacy policy is available at www.RaptorCares.com.

At this time, RaptorCares is available only to patients with cystinosis and caregivers in the U.S.

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Conference: Heroes Among Us (continued from page 1)


Ideally located in the very heart of Washington, D.C., Marriott Wardman Park boasts the combination of a relaxing luxury resort with quick access to many area attractions via an on-site Metro station. This is the perfect opportunity to visit the Smithsonian Museums, the National Zoo, and much more. The hotel is easily accessible by Metro from Washington airports and is a short walk from the National Children’s Zoo, theaters and restaurants.

Registration materials will be available in early 2013 on the CRN website and will also be mailed to families.
CRN Education & Awareness Update

by Jessica Jondle—Vice President, Education & Awareness

I am honored to be stepping in as the Cystinosis Research Network’s Vice President of Education & Awareness. I hope to continue the work done by Paula Shal during her time in the position, though I know I have enormous shoes to fill. I thank her for all she has done (and continues to do) to ensure that I am well supported in my new role.

A little about me—I am a 31-year-old with cystinosis, working full-time as an English and History teacher at a middle school in Berkeley, California. I teach six different subjects (one class each of ancient history, medieval history, U.S. history, and sixth, seventh, and eighth grade English) in addition to facilitating the school’s Student Council and Community Service programs, amounting to a school day without breaks and long evenings of preparation at home.

Though the hours are long, I continue—you could say that education is my passion. Nelson Mandela asserted that “Education is the most powerful weapon which you can use to change the world.” I agree wholeheartedly—when I step into my classroom and face my students, I hope that I am presenting them with the tools they need to grow into adults who facilitate positive change in their communities.

Likewise, to bring change to the cystinosis community—positive treatment advances, earlier diagnoses and medication access worldwide, and a new generation of patients who reach healthy adulthood and old age—education is our most powerful weapon. For that reason, the Education & Awareness Committee is a crucial part of the Cystinosis Research Network. By exhibiting at medical conferences, making scholarships available to graduating high school students looking to further their educational careers, and producing written materials to inform our community members (present and future), we further our goal of empowering individuals to create positive change.

When I was diagnosed with cystinosis in 1983, Internet research tools were unavailable. Yet I am where I am today because my parents educated themselves. From that very first library trip (during which they opened a large medical volume and discovered in the few lines devoted to cystinosis that my life might be limited to 10 years) to connecting with other families at conferences to always asking informed questions and keeping meticulous notes, my parents put me on a path to success and independence. I owe them a tremendous debt of gratitude for the opportunity I now have to educate others. I am passionate about treatment compliance (due to the result of my own noncompliance in my twenties) and spreading awareness about adult issues that the community has been necessarily silent on until recently. That there are many of us living as adults and more to come has necessitated changes in Education & Awareness. (We used to exhibit exclusively at pediatric nephrology conferences. Now we attend adult nephrology conferences as well.) What an exciting time in which we live!

Marybeth Krummenacker continues to represent us very well in NORD, making CRN a substantial presence in the rare disease community. Family Support and Education & Awareness committee members represented CRN at the recent ASN Kidney Week in San Diego, California. (See report on page 25.) I am blessed to be working with such a devoted group of people, and I know that together, we can change our cystinosis world.
Report from the National Organization for Rare Disorders (NORD) 

by Marybeth Krummenacker—CRN Board Member

It has been a very productive few months within the NORD and CRN communities and we continue to maintain a close relationship. CRN demonstrates its ongoing commitment to be a leader in the rare disease community. In addition to CRN attending the annual meeting and Gala held at the historic Union Station in Washington, D.C. in May, we continually are asked to participate in conference calls and meetings and are working closely with NORD and their staff. I attended the regional membership meeting in Philadelphia in September and these meetings have proven to be very well received by both member organizations and prospective new member organizations. Organizations are given an opportunity to network together and share information and listen and learn from some of the leaders of organizations describe what the ups and downs of the rare disease community can be and what the benefits are to sharing information to become active participants in the rare disease arena. Christy Greeley attended the DIA/NORD meeting this past October and was asked to participate in a press conference about what it is like to live every day with a rare disease like cystinosis and run a non-profit. The Drug Information Association (DIA), together with NORD, has sponsored this very successful meeting for the second time whereby representatives of the DIA, FDA, NIH, and advocacy organizations listen and network together to learn about the issues affecting everyone. In addition, José Morales was asked to speak on one of the panels to discuss the international relationship building within the rare disease community. All in all it was an opportunity to spotlight CRN and all that we continue to do as a rare disease advocacy organization.

I was scheduled to attend the NORD Board of Directors Retreat in November, however Hurricane Sandy decided to visit and our Board Meeting has been re-scheduled for January. With the coming of 2013 I will begin my second three-year term on NORD’s Board as well and it has been an honor to represent the cystinosis community. As we go to press we are awaiting the announcement of the 30th Anniversary Celebration of the signing of the Orphan Drug Act in May, and we await anxiously for updates being planned for Rare Disease Day on February 28, 2013.

CRN Adult Care Excellence (ACE) Initiative Update

Pediatric to Adult Care Transition Guide. The Adult Care Excellence (ACE) Initiative continues to work to provide resources for the cystinosis community. The Pediatric to Adult Care Transition Guide is available on the CRN website.

Cystinosis Standards of Care. We recently collaborated with Dr. Galina Nesterova and Dr. William Gahl at the National Institute of Health and developed the Cystinosis Standards of Care. We hope this resource will assist families and physicians in the medical care of individuals with cystinosis from infancy through adulthood. The Standards of Care are available on the CRN website.

Cystinosis Parent Handbook. The Cystinosis Parent Handbook will be available on the CRN website by December 1, 2012. The Handbook is a tool to use in a parent’s journey after a child’s diagnosis with cystinosis. Parents are likely filled with questions about cystinosis and their child’s future. The Handbook will help them get a better understanding of cystinosis and its treatment, and learn to care for their child and find support with others in the cystinosis community.
CRN Development Update

By Tim Miller—Vice President, Development

The New Kid.

In the middle of my sophomore year of high school my parents moved the family from Omaha, Nebraska to Lawrence, Kansas. The night before my first day of class I lay awake wondering what it would be like in a new school without a single friend to call my own. To say that I was a little anxious would be an understatement, but by the time my first class started I had decided to take this opportunity to reinvent myself. No one in the school knew my history. No one knew my shortcomings. I could be anyone I wanted to be if I just decided to.

By the end of my senior year I stood in front of the class of ’81 as a commencement speaker. I had tried out for a spot because I was nowhere near the valedictorian of my class. (And “nowhere near” the valedictorian is a gross understatement of my GPA!) But there I was, giving a speech I had written to 479 classmates who didn’t know I was alive two years earlier. I figured out how to reinvent my shy, intimidated, dyslexic self into a confident self-reliant young man.

So here I am, 32 years later. The new kid again. While more mature, with gray and noticeably less hair, I have decided to reinvent myself again. This time I’m joining a new school called Cystinosis Research Network. My little secret this time is I have no reason at all to be in this school. No one transferred me and I’m way too old to be learning anything new. I’m here because I want to be. I’m here because I want to help CRN raise money so they can do what they do best.

You see, I work for a company called C.H. Robinson. I started with them in 1989 after my wife and I graduated from the University of Kansas (Rock Chalk Jayhawk!). My journey with C.H. Robinson took me to Miami, then Kansas City, Wichita, and Salt Lake City, where I live now. In 2006 I asked my new office in Salt Lake City to select a charity to raise money for in an effort to give back to the community. While there where several foundations presented to the office, it was a business partner’s daughter who had cystinosis that touched our hearts. Since then, the office has held a tournament in June each year and has raised over $260,000 for CRN.

Earlier this year I offered to help CRN and volunteered for the Board of Directors. Well, one thing led to another and I was voted onto the Executive Committee as the V.P. of Development.

So what is the new kid’s plan, you might ask? There are two parts of it for me. First is passion. When I tell people that I’m involved with CRN they are amazed at the passion and desire I display to do something significant for my adopted cystinosis family. I want more of the CRN family to feel the passion I have to help this organization provide help to its members. CRN plays a critical role with support and research. CRN’s value cannot be
equated into dollars and cents when a family needs to know how to deal with their child’s first kidney transplant. At the same time, having a support system and providing money for research does cost real money. So my first plan is to find those passionate people who understand that a life without CRN would be crippling.

My next plan has two parts. The first is simple. It’s a call to you all to do something. Take action. Whether it’s a letter-writing campaign, a fundraiser at your local Buffalo Wild Wings, or a fun walk, do something and give back to CRN so it’s a healthy network. I have plenty of ideas and have had several share their ideas with me. I took this position to make a difference. I, the Executive Committee, and Board members will help you. You just need to let us know you are willing. We will help you create an action plan and provide support for you. You are not alone, but we can’t do everything. Your involvement and actions are critical for this volunteer organization.

The second side of my plan is to implement a large event that will create awareness for cystinosis while raising thousands of dollars for CRN. The world is full of passionate people who love to golf, play tennis, run, bicycle, and ski. The point is, these people all participate in events so they can enjoy their “passion.” Whether it is a 5K run or a ski day in the Rockies, the goal of the event is usually to raise money for a charity, foundation, or special need. I see a bike or running event in the future for CRN. What I need right now is help. A Project like this needs more than just good intentions. It needs an active group of people who will carry the torch forward and build upon successes.

So here is my request. It’s simple and straightforward. If you would like to help the new kid out in raising funds for the future of CRN, then take the first step and send me an email. In the email tell me a little about yourself and the type of commitment you would be willing to make and be passionate about. If all you have time for is putting addresses together of your friends, coworkers, and family for a letter campaign, I know the Board can help you create a letter. If you love to bike and can see yourself starting the first annual “Ride for Cystinosis,” then let me know. All I ask is that you find your level of commitment and passion and let the new kid help you—together we’ll have an impact.

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

Visit [http://www.cystinosis.org/families-in-your-area](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!
6th Annual C.H. Robinson Golf Event Raises $79,400 for CRN

by Tim Miller—Vice President, Development

I was a little disappointed this year at the 6th Annual C.H. Robinson Charity Golf event for cystinosis. It was a sunny, 71-degree day with a light breeze, but everyone knows 72 degrees is the perfect temperature for a Charity event! But it was not a total loss...We did manage to raise $79,400.

With the help of 33 C.H. Robinson offices that Co-Sponsored the event or Sponsored a hole or green, this event did raise the bar a little higher in 2012. But I can’t forget all the families, friends, and customers who have returned to Utah for this event year after year. Jeanne Oki has been a Co-Sponsor of this event for years and always brings a team of 8 to enjoy the day. Our friends at SRS are the first to renew their annual commitment as a Hole Sponsor. Northwestern Mutual, PMI, and the Vanguard teams joined despite the questionable weather forecast.

This year Jeff Larimore attended the event to voice his gratitude for the funds this event raises for CRN. He also got his first taste of the unique rules that are created to make this a fun and successful event. Some of those rules include 30 inch licorice rope to finish out a shot or a sling shot to advance your ball with no stroke penalty. Doubling raffle ticket and mulligan balls are just other ways to keep the day fun while focusing in on gathering more donations for CRN. I think Jeff enjoyed the day but teared up a little as he spoke to the 200 in attendance and reflected on the impact that CRN has on his family.

Mason Reed made the event this year and took an opportunity to “tee off” for a $5 donation on the “lefty righty / righty lefty” hole. That’s the hole where participants had to tee off using a left handed club if they were right handed or vice versa. While I’m not sure how this little salesman managed to convince golfers to make a donation for his tee shot, I do know Mason was a “big hit.” This just so happened to be the hole sponsored in Mason’s name and I hope he will return in 2013 for a repeat performance.

I know there are many others from CRN that Pam and Tahnie Woodward invited to the event. Attendees included Frankie and Laura McGinnis; Jessica and Wayne Jondle; Ryan, Kylene, Max, and Deb Reed with Mason and Jaxon; and Autumn and Aaron Goodridge with Felicity. I’m grateful for all the support you showed by coming in spite of the dismal weather conditions. I’m hoping the
2013 event on June 8th is just as disappointing and raises $100,000...or more.

I want to thank C.H. Robinson World Wide for its continued financial support of this event, Thanksgiving Point Golf Course for their amazing staff, my friends who amaze me with support, my staff who keep giving, and my family that fills my life with meaning.

If you would like to know about this golf event to build your own CRN fundraiser or create your own unique fundraiser, please contact Tim Miller at tmiller@cystinosis.org.

CRN Exhibits at the American Society of Nephrology’s Kidney Week in San Diego

by Jessica Jondle—Vice President, Education & Awareness

CRN current and former Board members Pam Woodward, Frankie McGinnis, Karen Gledhill, and Jessica Jondle attended the ASN’s premier nephrology meeting, Kidney Week, at the San Diego Convention Center October 30 - November 4, 2012. This event featured presentations by the world’s leading doctors and researchers in the field of nephrology and was attended by thousands of healthcare professionals. Cystinosis was highlighted in four moderated talks on November 1: Cystinosis: Natural History and the Impact of Therapy by CRN Scientific Review Board Chairperson Dr. Bill Gahl; CTNS Gene: Discovery and Beyond by Dr. Corinne Antignac; Transcriptional and Posttranscriptional Regulation of the CTNS Gene by Dr. Francesco Emma; and Approaches to the Genetic Rescue of Cystinosis by Dr. Stephanie Cherqui.

The CRN booth provided information to nephrologists worldwide who were eager to know more about cystinosis. Many had one or two cystinosis patients, but some were relatively unfamiliar with the disease. CRN’s presence at these meetings is hugely impactful in raising the kind of awareness that is so necessary for early diagnosis and proper treatment.

Dr. Jerry Schneider stopped by our booth and obliged us with a photo. Also present were CRN Scientific Review Board Member Dr. Craig Langman, who had a poster presentation regarding RP103/Procysbi Phase 4 study data; CRN Scientific Review Board Member and adult nephrologist Dr. Roslyn Mannon; and Drs. Paul Goodyer, Julie Ingelfinger, and Ewa Elenberg, all CRN Medical Advisory Committee members. We also spoke with Dr. Elena Levchenko, who had a poster presentation regarding copper levels in patients on cysteamine therapy.
My parents, childhood sweethearts, married during the post-WWII euphoria. They had both served their country honorably—my mother as a member of the Army Nurse Corps and my father as a blimp pilot in the Navy. (How’s that for a unique assignment?) Dad loved the Navy and might have made it a career had the Korean Conflict not started right as he found out that my mother was expecting their first child. Not even an offer of a bump up the officer’s ladder could entice him to miss the big event.

On November 25, 1952—just two days before Thanksgiving—my gorgeous, red-headed big brother was born. My parents were over the moon! They named him Patrick Joseph after my grandfather and, since my father was also named Patrick, they called him “Patty.” Not only did they finally have an heir to carry on the proud O’Shaughnessy name, but my brother was the first male born amidst dozens of women on my mother’s side of the family.

Everything seemed to be perfect—that is until the day that my mother told my father that their beautiful infant son was dying. Can you imagine the shock? No, she hadn’t seen a doctor and, to my father, Patty looked perfectly healthy, but my brilliant R.N. mother knew that there was something terribly wrong. With that announcement, she began her odyssey of going from doctor to doctor stating her claim of impending doom only to have the doctors—and my father—determine that she was utterly nuts. Sure, Patty was a little small for his age and he wasn’t the greatest eater, but they really didn’t see much else wrong.

When Patty was almost three, I was born a month premature. Even so, I was a robust baby and my parents immediately took me home as a proud addition to the family. It wasn’t long until I looked like a mutant compared to my small brother. My favorite photo shows him giggling while trying to hold me. As I rapidly grew, it became even more apparent to my mother that my brother was failing to thrive. While I gobbled everything that came within the proximity of my mouth, Patty often refused to eat at all. When he did, he often vomited.

Over the course of the next year, doctors began to take notice of Patty’s small stature, the rickets-like deformity in his legs, and some sort of crystals in his eyes that made them sensitive to light, but none seemed to have a clue about what was at the root of his problem. Meanwhile, my mother became pregnant again, giving birth to my sister, Maureen, just one year and a week after having me. She was like a tiny, blonde-headed doll and my parents were 60% of the way toward achieving their “ideal Catholic family” goal of five children.

It did not take my mother long to sense that my sister was gravely ill with whatever my brother was suffering from and so the visits to various doctors continued, only now my mother was claiming to have two dying children. The physical signs were there, but my father did not want to see them. You can imagine what kind of effect the “Chicken Little Syndrome” was having on my parent’s marriage and I was literally becoming the forgotten child who became increasingly difficult to handle just to get noticed.

In 1957, my mother took all three of us to see an incredibly gifted physician by the
name of Ira Rosenthal who practiced at the Illinois Research Hospital in Chicago. He recommended that my brother undergo exploratory surgery so his organs could be examined visually. Patty spent almost a year in the hospital before Dr. Rosenthal properly diagnosed him—and Maureen—with an incredibly rare genetic disorder known as cystinosis. He further told my parents that, with a three out of four possibility of passing the disease on to another child, they should not have any more children and be glad that they had one “normal” child.

Because dialysis was still a very new technology, it was not being offered to patients without the expectation of complete recovery and kidney transplantation was still very experimental. So, it was determined that the best course of treatment for my siblings would be to have my mother make a medicine that consisted primarily of potassium acetate and to treat the remainder of their symptoms with comfort measures. Dr. Rosenthal felt that my brother was already on borrowed time, at the age of six, and that my sister might live to be his age if she were lucky. However, he greatly underestimated my mother’s nursing skills and devotion to her children.

I intend to write a book that details both the trials and fascinating miracles we experienced because of the privilege of having Patty and Maureen as beloved members of our family; however, in the interest of brevity, suffice to say that our lives were different than those of other families. For instance, our refrigerator and broom closet were stocked with bottles and bottles of electrolyte replacement fluids. There were wax paper bags and empty cardboard milk cartons in liberal supply around the house and in our cars in case the kids suddenly succumbed to their constant nausea. And, to this day, the smell of a pine cleaner immediately reminds me of the bucket of Lestoil that sat on the top step of the basement steps so any mess could be cleaned up as soon as it happened. That bucket of pine water needed to be changed at least daily and sometimes even more often.

In the end, my beautiful, serene little sister, who never grew taller than the middle of my chest, died relatively peacefully of complications from a heart attack just four days before her ninth birthday. I couldn’t help but feel that Patty watched her funeral proceedings as if they were foreshadowing his own. Even though I was only ten years old, the look on his face broke my heart. Just six months later, my courageous big brother died a horrific death due to uremic poisoning after his kidneys completely failed. He spent six awful weeks in the hospital before he finally reached out between the bars of his bed, where he lay swollen almost beyond recognition, to try to comfort me when I came to say goodbye. That was Patty in a nutshell and almost 50 years after his death, friends still write tributes on Facebook about how he influenced their lives for the better.

Dr. Rosenthal credited my mother with the quality and longevity of Patty’s and Maureen’s lives. Mom filled our home with laughter and joy while she did her very best to minimize all suffering. She made sure that they lived their lives to the fullest. We vacationed all over the eastern U.S., rode horses, fished, swam in our own pool, and did way more than most healthy kids ever got to do in such a short period of time. As their final act of love, my parents made the heroic, but unpopular decision in the eyes of the church, to donate both my brother’s and sister’s organs for further study in the hope that something could be learned to spare another family such tremendous heartache. I don’t know if anything good ever became of that, but I know that it helped them to feel better in a tiny way.

I’d like to be able to say that my family took their cue from Patty and moved on with courage and determination, but losing two children six months apart pretty much destroyed my parents and their marriage. I, however, had already built a strong faith in God from attending church with my brother, and so I healed to a much greater degree than my parents ever did. In fact, I worked as a hospice volunteer and in home health caring for the terminally ill—including children. I couldn’t bring healing, but I hope that I was able to be of support and comfort while honoring the memory of Patty and Maureen with my care, empathy, and compassion. I have also spent 20 years in pastoral ministries and we, in church work, have adopted a saying, “Life ain’t for wimps.” I think that is exponentially true for families dealing with sick children, but I am so encouraged to know that there is tremendous hope for those diagnosed with cystinosis. When I write that book, assuming anyone ever buys it, I want the proceeds to go toward finally finding the cure. With all the advances that have been made, I believe that with more research dollars, that goal can be achieved in my lifetime. May it be so!
Haunted House Raises Funds for CRN

Sakats Manor Haunted House is a group of individuals consisting of family and friends who decided to take their love for Halloween to a whole new level. Halloween 2012 marked the group’s fifth year.

In 2010, Drummer Boy Campground agreed to donate an entire site to our crew, making for the biggest haunt yet. A large amount of our building materials were also donated by the Campground. At this point, we officially became Sakats Manor (SAKATS represents the creators—Sean, Alisha, Karen, Alex, Tara, and Shannon). This is the year when we tripled in size. We spent three days erecting a 20X36 foot building, complete with six scare rooms, a front porch, a decorated graveyard, and numerous additions. After an additional day to add the roof, and about six weeks of decorating, Sakats Manor successfully brought in over 2500 visitors. We collaborated and decided to forward our donations to a cause near and dear to us, the Cystinosis Research Network, in honor of Laura McGinnis, one of our very own on Crew 165.

Thanks to friends and family of Crew 165, we continue to grow and support our cause. This year, we offered yet another fantastic season of haunts and fun for all, while at the same time supporting our cause and our love for Halloween. Our project has grown in leaps and bounds, and we hope to continue to pass our word along for our cause. At Sakats Manor, Crew 165 “scares because we care.”

Who is our motivation? In the words of Frankie McGinnis, Laura’s mother: "Laura Kathryn McGinnis came roaring into this life on March 23, 1996, 8lbs. 15.5 oz. of screaming femininity. At birth, Laura was perfect. Perfect lasted for us a total of two weeks. This was when we had our first ER visit and our first specialist referral, to a gastrointestinal physician. Laura was having respiratory issues, spiking fevers of 103, and vomiting frequently. The doctor suspected Laura had cystic fibrosis. He placed her on two medications to help her food move quicker out of her stomach and to help with reflux. Laura was given every test known to man for eating and respiratory issues; her symptoms were treated, yet she continued to get sicker. Her size became more markedly delayed every day. She was hospitalized at 11 months for a g-tube placement to try to get more nutrition to her; this was a simple procedure which we expected to take place in same-day surgery. Our one day ended up being a month. Laura did not react well to the withholding of fluids and anesthesia, and as her rollercoaster labs continued someone mentioned she should be seen by a nephrologist. Dr. Wm. Lane Robson came into our room looked at Laura, looked at her massive medical chart and informed us he was ordering a blood test but he was 99% sure our daughter had cystinosis. We had no idea what this man was talking about but after a quick search of the medical library and seeing the words Always fatal, we knew he had to be wrong. He wasn’t. Dr. Robson sat me down and handed me a notebook of every article ever published at that point on cystinosis. He informed me it was my duty as Laura’s mother to be her advocate, learn as much as possible, fight harder than I ever imagined, and learn to get every available advantage for Laura to see that she had the best life possible. He did not give me a moment for woe is me; he would not allow any pity—only an empowerment for which I will be eternally grateful.
Laura's first few years after diagnosis were rough. She averaged a 2lb weight gain for her first five years, which if you do the math, leaves for one tiny little person. Fanconi's was extremely hard to control and we had to get creative to get her into some sort of normal range. She had no "reserve" and adding in her asthma made for many hospital trips. Vomit was a way of life for us. To this day, we still keep gallon zip-lock bags in our cars—because you just never know. Two summers ago Laura had one leg operated on to help with her "knocked" knees; she has responded well. Her g-tube was removed when she was 12 and the look on her face when she saw her stomach without a tube is one of the most precious moments of my life; she rubbed it just like she had as a baby. It still brings a tear to my eye thinking of how beautiful she thought her stomach was. We tried growth hormone on two separate occasions but for Laura it was not worth the trauma. She is needle phobic, I say that with a grin since you can prod her veins all day, but try and give her a shot and she goes ballistic. Nothing will persuade her to willingly get a shot of and we are firm believers in picking your battles. Laura is well aware that she is going to be a small woman. She does not care. She is 4' 8"ish, 80lbs, and quite happy with herself.

A day in the life of Laura depends on the day; no two are ever the same. She is on intermittent homebound service, which means she goes to school when she feels up to it; otherwise she has a teacher come to the house who brings her assignments and helps her as needed. She has a restricted license which allows her to drive alone until 6pm, which has done wonders for her self-esteem and both of our mental health since we get time away from each other. She dances competitively in jazz and hip hop which she loves. Ours may not be completely typical, but we have made a happy life and fill it with as much love and joy as we can."
Family Conference
Sponsorship Opportunities

CRN is pleased to announce that the 2013 Family Conference will be held July 18-20, 2013 at the Marriott Wardman Park Resort in Washington, D.C. This will be CRN's sixth family conference.

We cordially invite you to take advantage of the great sponsorship opportunities available to not only celebrate the remarkable accomplishments of the Cystinosis Research Network, but to support the vision of CRN: The discovery of improved treatments and ultimately a cure for cystinosis. Our commitment to our community lies in our tireless efforts to provide family assistance through facilitating several support groups as well as hosting a biennial conference. At this conference we bring families and the medical community together in hopes to accelerate the accomplishment of our stated goals and objectives.

Past sponsors include:

CRN SPONSORSHIP LEVELS

Platinum Event Sponsor $30,000
- Dinner invitation to board and medical advisory meeting/dinner
- Premium placements in donor banner displayed at conference
- Verbal recognition at the conference opening
- Verbal recognition at the cocktail reception
- 11x17 laminated color poster placed at the event
- Logo on website with direct link to donor
- Logo and/or advertisement in pre-conference newsletters
- Included in email blasts and all conference related announcements and materials (time permitting)
- Top Logo representation on conference t-shirt
- Donor literature included in each guest room
Silver Event Sponsor $15,000
- Dinner invitation to board and medical advisory dinner
- Verbal recognition at the conference opening
- Verbal recognition at the cocktail reception
- 11x17 laminated color poster placed at the event
- Logo on website with direct link to donor
- Logo in pre-conference newsletters
- Included in email blasts and all conference related materials (time permitting)
- Logo representation on conference t-shirt
- Placement in donor banner displayed at the event

Bronze Event Sponsor $10,000
- Verbal recognition at opening reception
- Verbal recognition at cocktail reception
- 11x17 laminated color poster placed at the event
- Logo on website with direct link to donor
- Logo in pre-conference newsletters
- Included in email blasts and all conference related materials (time permitting)
- Logo representation on conference t-shirt
- Placement in donor banner displayed at the event

Breakfast Sponsor $3000
- Recognition at conference opening
- 11x17 laminated color poster placed at each day’s breakfast
- Placement in donor banner displayed at event
- Included in all the event’s email announcements
- Logo on the website with direct link to donor

Lunch Sponsor $3000
- Recognition at conference opening
- 11x17 laminated color poster placed at each day’s breakfast
- Placement in donor banner displayed at event
- Included in all the events email announcements
- Logo on website with direct link to donor

Napkin Sponsor $1000
- Donor’s logo printed on cocktail napkins displayed at the cocktail reception
- Placement in donor banner displayed at event
- Included in all the event’s email announcements
- Logo on website with direct link to donor

Registration Table Sponsor $1000
- 11x17 laminated color poster placed at the registration table
- Placement in donor banner displayed at the event
- Included in all the events email announcements
- Logo on website with direct link to donor

Inspirational Speaker Sponsor $500
Cocktail Reception Sponsor $500
Conference Materials Sponsor $500
Childcare Sponsor $500
Closing Dinner Sponsor $500
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 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 Cystinosis Research Network today!

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

Pam Woodward
VP Family Support, Cystinosis Research Network
Join the Cystinosis Research Network Today!

<table>
<thead>
<tr>
<th>Category</th>
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International: (Including Canada) Base rate (see above categories) plus $10.00 for postage.
Payable in US dollars

Please complete the form & mail with check payable to CRN to:
Cystinosis Research Network
302 Whytegate Ct.
Lake Forest, IL 60045

***************************************************************************
Name_________________________________________________________________________
Street________________________________________________________________________
City & State_______________________________________ Zip Code__________ Country ____
Phone__________________________Fax_______________________Email________________

Join A CRN Support Group

Looking for a way to communicate with others in the cystinosis community on a day-to-day basis?

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

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

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

To join a support group, visit www.cystinosis.org.
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 Web with GoodSearch & Raise Money for CRN

Every time you use GoodSearch.com to search the Web, a donation is made to the Cystinosis Research Network! GoodSearch.com is powered by Yahoo!, so you get the same 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 goodsearch.com and select Cystinosis Research Network where it says "Choose your cause." Then search like you normally would!

The more people who use this site for CRN, the more money is earned. So please tell your friends and family!

Make Purchases at GoodShop & 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, go to goodshop.com, choose CRN as your charity, and 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, American Eagle Outfitters, and 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.

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<thead>
<tr>
<th>Agency Name</th>
<th>The Cystinosis Research Network, Inc.</th>
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<tr>
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<td>04-3323789</td>
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<tr>
<td>Address</td>
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</tr>
<tr>
<td>Telephone</td>
<td>1-866-276-3669 (toll free), 1-847-735-0471</td>
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<td>Fax</td>
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</tr>
<tr>
<td>Web Page</td>
<td><a href="http://www.cystinosis.org">www.cystinosis.org</a></td>
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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.