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2013 Cystinosis Research Network Family Conference - A Great Success!

The 2013 Cystinosis Research Network (CRN) Family Conference was held July 18 – 20, 2013 at the Washington Marriott Wardman Park Hotel in Washington, DC. Approximately 300 family members, physicians, researchers, and volunteers attended. Included in the attendance were 14 families who had never attended a family conference before. All attendees were able to learn about the latest updates in cystinosis research through lectures, workshops, poster sessions, and medical and patient panels. There were many opportunities to socialize with other patients and families, which provided valuable connections and friendships that will last a lifetime. **(See Family Conference, page 9)**



Children and adults living with cystinosis attending the 2013 CRN Family Conference in Washington, D.C., July 18th to 20th.

CRN Announces 2013 Scholarship Recipients

The Cystinosis Research Network is pleased to announce the recipients of our two 2013 CRN \$1,000 scholarships. After careful consideration of six excellent and qualified candidates, the CRN Scholarship committee has selected, Elizabeth Patterson, to receive the CRN Academic Scholarship for an individual with cystinosis and, Kole Binger, to receive the CRN Sierra Woodward Sibling Scholarship. **(See Scholarships, page 4)**

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Message from the President Jeff Larimore

Perseverance Leads to a Productive Year

The responsibility of writing the President's letter provides the opportunity to share the ongoing successes of many years of research, recap the current progress of expanding awareness of Cystinosis to families and the medical community, and to set an ongoing agenda where the mission and vision of the Cystinosis Research Network is realized. We are pleased to present throughout the Fall/Winter edition of the *Cystinosis Advocate* many exemplary stories which portray these significant achievements.

Year 2013 has been the highlight for many throughout the Cystinosis community. After years of barriers and challenges, Sigma Tau Pharmaceuticals and Raptor Pharmaceuticals received FDA approval for their reformulation of cysteamine bitartrate which provides Cystinosis patients with options for improved treatment. The Cystinosis Research Network and its members have and will continue to support the tireless work in the study of Cystinosis that these two organizations provide.

In May, the National Organization of Rare Diseases (NORD) recognized the years of "heroic" commitment to the study of Cystinosis to Dr. Jess Thoene and Sigma Tau Pharmaceuticals. Representatives from CRN were attendance and were applauded as Dr. Thoene graciously commended CRN for its strong support as a patient advocacy organization.

The 2013 Family Conference held in July brought together the largest number of families and attendees to a CRN Conference. The topics of discussion were broad, diverse and engaging. The Conference brought together 24 renowned medical professionals in one sitting to discuss recent developments in Cystinosis and the current progress in laboratory research and clinical study with sites on an eventual end to Cystinosis.

How do the achievements of the past year relate to the present time? The commercial production of Sigma Tau's Cystaran has replaced the challenges families and patients faced with compounding. The availability of Raptor's Procysbi provides a 12-hour dosing schedule that allows patients and caregivers the opportunity for a full night's sleep. Delivery of each product is now to the front door –"you gotta love logistics!" Many within the CRN community have commented personally and through social media that transition to these medications have provided enhancement in the quality of life for patients.

The transition to the reformulated medications has not been without difficulty for some patients. Research and development continues on both products so that the best treatments can be available to the community. CRN will continue to request and support patient feedback so that all areas of concern can be addressed.

It is fairly easy to state that perseverance has been a key building block for the most recent accomplishments within the Cystinosis community. The Cystinosis Research Network has been fully engaged and shares the joy in the triumphs but will not shy away from addressing the ongoing questions that should be asked.

What will drive the Cystinosis Research Network forward? Initially, it is our sincere appreciation for the donations and grants received. CRN remains an all-volunteer advocacy group and a core belief is the importance of being proper stewards of these investments.



Jeff Larimore

Areas of concentration for CRN in the upcoming year are:

- Enhancing communication on the full scope of managing Cystinosis with newly diagnosed families,
- Working actively within the Cystinosis community to inform patients and caregivers of the broad based resources available through CRN's website and/or professional network,
- Expanding awareness of Cystinosis beyond pediatric circles to assist in the transition to adult care,
- Presenting educational forums that address current developments in the area of Cystinosis along with other topics that are instrumental in patient development,
- Apply resources to areas of research and development that concentrate on achieving the mission of CRN,
- To hold 1-2 fundraising events per month and/or secure a material corporate grant over the same period,
- To inform persons managing Cystinosis that volunteer work within the community pays dividends in their professional endeavors,
- Being proud of the work accomplished and to be a standard bearer for the Cystinosis community.

On behalf of the Board of Directors of the Cystinosis Research Network, thank you for your continued support and all the best wishes for this holiday season.

Cheers,

Jeff Larimore, President

CRN Board Welcomes New Member, Patty Blais

At the 2013 Family Conference held in Washington, D.C. in July, the CRN Board met in part, to elect the 2013-2014 Board of Directors. Several current Board members were appointed to new positions on the Executive Committee. Current Board members assuming new positions are Christy Greeley who along with her job as Executive Director will also be Vice President of Research, Terri Schleuder who was elected as Vice President of Education and Awareness, and Deb Reed who assumed the job of Secretary. Jeff Larimore, Jose' Morales, and Jenni Sexstone will stay on as President, Vice President of Development and Treasurer respectively. In addition the Board welcomed Patty Blais to become its newest member. **(Please read more about Patty on page 4.)**

Scholarship Recipients (continued from page 1)

Elizabeth is an English major continuing her studies at Colorado State University with plans to pursue either an M.A in nonfiction or an M.F.A. in poetry. Her poetry was recently published in Colorado State University's literary journal, *The Greyrock Review*.

Kole graduated from Verona Area High School in Madison, Wisconsin, and plans to begin her college career at the University of Wisconsin-Madison following a premed academic path. In addition to having a brother with cystinosis, Kole, has battled her own serious health challenges over the past year and a half. In spite of this, Kole, has assumed various leadership roles at her school while maintaining an excellent GPA.

We wish both of these young women the very best in the future as they pursue their academic and personal goals.



Kole Binger

A Thank You Message from Kole

I am so unbelievably honored that I was lucky enough to be given this opportunity. Ever since I have been old enough to comprehend what Cystinosis is, I have been driven to someday do my part to help to find a cure. I have always been interested in different medical-related issues, however I hold Cystinosis near and dear to my heart. It is one of my personal goals to be able to be a part of the process in finding a cure for this disease. I am majoring in Pre-Med, and as many of you know, 12 years of schooling can add up. This scholarship will be able to help my parents and I with the tuition, and also help us to save money so I can one day go to medical school and accomplish my dreams! Thank you so much!



Patty Blais

Patty Blais Joins CRN's Board of Directors (continued from page 3)

Patty Blais worked in retail management until 1997 when she decided to become a stay at home mom. In 2010 she returned to college to earn a degree in Legal and Office Administration. She is one semester away from completing that degree. She was recently hired as a customer service representative for NG & G beginning her reentry into the work force. Her grandson, Owen, was diagnosed with Cystinosis in September 2012. She and her family worked together form a board of ten people to form Owen's Outlook; a non-profit organization to benefit CRN. They are working toward becoming 501c3 certified and have completed their 2nd event...a golf tournament this past September.

Utter May!Hem in Maryland Raises Money for CRN

by Julie Ayers

The streets of Baltimore were filled with music on May 11th in an effort to raise money for cystinosis research and to assist those living with the genetic disorder. Well, it was really just one yard in Timonium, Maryland, a suburb of Baltimore, filled with music, but the bands were so great that I'm sure all of Baltimore wished they could have been in attendance, having fun, and doing good with the Ayers & Mutchler family.

This is the second year our family has hosted the **May!Hem** music festival fundraiser to benefit the Cystinosis Research Network.

The idea for **May!Hem** developed after I won a performance by a rock band for \$60 in an auction fundraiser at my church. Not sure when and how to use this band, I decided to hold a party in our backyard in May to celebrate a bevy of birthdays in a grand fashion. As plans evolved, we included the band I won in the auction, and several other bands, and morphed the event to be not only a May-Birthday-Bash-Celebration, but also a fundraiser for CRN. Guests attending were asked to consider making a donation. The first year, our daughter, Sierra (with cystinosis), set up a lemonade stand the day of the event and we also put out a money box into which people could voluntarily drop some bills, checks, or change. We raised about \$1400 at the first **May!Hem**.



Musicians raising money for CRN at May!Hem in Maryland



Frankie and Laura McGinnis, Julie Ayers and Pam Woodward enjoy the May!hem fun!

This year, we put a bit more thought into how to generate additional funds, so charged admission, added on online auction of items and services from local businesses, held a 50/50 raffle at the event itself, and asked people to consider making donations beyond the amount requested for admission. **May!Hem: The Sequel** raised about \$3000. Yay for doubling!

All the bands & musicians (Dog Day Afternoon, The Fifty 7's, Bald Mountain Band, Paul Azzam & Lelah Mahoney) donated their services for **May!Hem: The Sequel**. The event also included belly dancing, a magician, and a poetry reading. Friends donated auction items, promotional material development services, tents, and helped plan

and run the event. I'm so grateful for all of the support, the funds raised, and especially the FUN raised by **May!Hem!** Next year: **May!Hem Revisited?!** Can we double our double?



CRN Attends NORD 30th Anniversary Celebration Honoring Rare Disease Heroes

Press Release

Heroes of the rare disease community joined future advocates on the stage at the glittering 30th Anniversary Celebration of the National Organization for Rare Disorders (NORD) on May 14th at the Mellon Auditorium in Washington DC. CRN board members Christy Greeley, Marybeth Krummenacker, Jeff Larimore, and Jose' Morales were in attendance.

Before an audience of 500 people, Representative Henry Waxman (CA) hailed Abbey Meyers and other patient advocates for their leadership and vision in promoting federal incentives to encourage the development of treatments for people with rare diseases. Representative Waxman, the author and chief sponsor of the *Orphan Drug Act*, was one of the honorees for the event, which was a highlight of NORD's year-long 30th Anniversary observance.

On an evening that many are describing as "magical", three young representatives of the next generation of rare disease patient advocates also spoke. Megan Barron, a recent Duke University graduate; 11-year-old Brooke Foster; and 8-year-old Ian Brown talked about the rare diseases of particular concern to them – epidermolysis bullosa, mastocytosis, and Langerhans cell histiocytosis – and thanked the rare disease heroes for the work they have done on behalf of patients and their families.

In addition, by three patients and the parent of a patient -- Kathryn Bundy, Christian Jacobs, Reina Weiner, and Marybeth Krummenacker, Cystinosis Research Network board member – presented awards to senior officials from four companies for innovative products approved by the Food and Drug Administration in 2012.

U.S. Deputy Surgeon General Boris Lushniak, MD, served as guest emcee, assisted by longtime NORD adviser and board member Frank Sasinowski. Entertainment was provided by the Howard University Jazz Ensemble and American Idol finalist Siobhan Magnus.

The 30th Anniversary NORD Honorees are:

FOR LEADERSHIP IN PUBLIC POLICY:

- William V. Corr, who served as Counsel to a Subcommittee chaired by Rep. Waxman during the Orphan Drug Act hearings and currently is Deputy Secretary of Health & Human Services
- Senator Nancy Kassebaum (KS-retired)
- Representative Henry A. Waxman (CT)



Dr. William Gahl with his wife Mary, and Dr. Jess Thoene with his wife Marijim, at the NORD 30th Anniversary Celebration.



Jessica, Sarah and Katie Larimore

FOR VISION ON BEHALF OF PATIENTS:

- Stephen C. Groft, PharmD, Director of the NIH Office of Rare Diseases Research
- Marlene Haffner, MD, MPH, Director (retired) of the FDA Office of Orphan Products development
- Lars-Uno Larsson, founder of Swedish Orphan International AB and early industry advocate for orphan product development
- Abbey S. Meyers, advocate for the Orphan Drug Act and President Emeritus of NORD
- Jess G. Thoene, MD, cystinosis expert and CRN Scientific Review Board member and researcher, advocate, and one of NORD’s founders and original medical advisers

FOR INNOVATIVE ORPHAN PRODUCTS APPROVED IN 2012:

- Aegerion, Inc., for Juxtapid for homozygous familial hypercholesterolemia
- NPS Pharmaceuticals, Inc., for Gattex for short bowel syndrome
- Onyx Pharmaceuticals, Inc., for Kyprolis for multiple myeloma
- Sigma-Tau Pharmaceuticals, Inc., for Cystaran eyedrops for cystinosis (award presented by Marybeth Krummenacker)



Jeff Larimore, Marybeth Krummenacker, Christy Greeley, and Jose’ Morales represent CRN at the NORD Gala.

Marybeth Krummenacker Presents Award to Sigma Tau For Development of Cystaran Eye Drops

by Marybeth Krummenacker

As we move towards the end of the year and we look back over the year that the Cystinosis community has had, there is only one word that comes to mind.....extraordinary! And I have come to realize even more the role that NORD has played in so much of what has happened in our little rare disease community and the honor I have to be on their Board of Directors.

The Gala held in Washington D.C. in May celebrating the 30th anniversary of the signing of the 1983 Orphan Drug Act was extraordinary. Without that significant piece of legislation, the Cystinosis community would have NEVER gotten approval of cystagon.....cystagon, is #41 on the list of the first 100 drugs approved as a direct result of the Orphan Drug Act of 1983. At the Gala, I had the distinct honor to present an award to Sigma Tau Pharmaceuticals for their unyielding commitment to the Cystinosis community. Sigma Tau stood by the side of the community for over 20 years and has remained committed to providing this eye drop therapy which makes a huge difference in the quality of life of patients. **(Continued on page 17)**



Marybeth Krummenacker presents an award to Dave Lemus from Sigma Tau Pharmaceuticals for the company’s dedication to the Cystinosis community by bringing Cystaran eye drops to market.



Erik Pekli

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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!

2013 CRN Family Conference A Great Success! (continued from page 1)

The conference theme, "Heroes Among Us" was exhibited through the conference by our speakers, professionals, and volunteers who all went the extra mile to ensure the conference experience was a positive one for all. Families had the opportunity to nominate their "Heroes Among Us" and posters of the Heroes were displayed throughout the conference. Most notably, CRN recognized Dr. Frederick Kaskel with the "Above and Beyond" Achievement Award. Dr. Kaskel was recognized for his extraordinary and on-going efforts to improve the lives of everyone in the cystinosis community. We are grateful to have Dr. Kaskel engaged in our mission.



Above and Beyond Achievement Award, presented to Dr. Dr. Fredrick Kaskel, by Marybeth and Laura Kruppenacker, and

With the support of our generous sponsors, CRN was able to provide three days of programming which was aimed at both the families and professionals. Wednesday began with the CRN Board of Directors meeting. The conference registration began on Thursday. CRN Scientific Review Board also met on Thursday to review scientific strategies. Families were welcomed to the conference on Thursday evening with a party with delicious food and entertainment by "Batman" and a face painter. Families had an opportunity to share their journeys with one another on Thursday evening during the Family Introductions Ses-



Medical panel of cystinosis experts answering questions from attendees.

session. Highlights of Friday's program included presentations about gene therapy and stem cell treatments for cystinosis. Experts also presented about how cystinosis affects the body in many ways including the kidney, neurological, endocrine, pulmonary, and gastrointestinal issues. The day concluded with the medical panel of 24 cystinosis experts who answered questions from the audience about a wide variety of issues related to cystinosis. **(continued on page 10)**



Shea and Colleen Hammond with Cheri and Morgan Friend at the Welcome Reception Thursday night.

Family Conference (continued from page 9)

On Saturday, updates about Cystaran were provided by Sigma Tau Pharmaceuticals and Raptor Pharmaceuticals provided an update regarding Procsybi. Highlights of Saturday included the "Living with Cystinosis Panel" presentation where teens and adults living with cystinosis shared how they live their lives to their full potential given the challenges they face. Discussions about cystinosis in Emerging Adulthood and Strategies of Successful Families were also highlights of the conference. A presentation detailing the importance of effectively coping with stress was helpful for families and individuals living with cystinosis. The afternoon workshop sessions allowed attendees the opportunity to interact with the experts on an in depth level. Finally, the highlight of the conference may have been a farewell dinner dance – where we were able to celebrate our time together with our cystinosis family.



Vera Toth-Fejel And Patrick Reichenberger from Raptor Pharmaceuticals provided information and updates on Raptor Care and Procsybi.



Lesli King representing Sigma Tau Pharmaceuticals

Childcare was exceptional and included a teen lounge, swimming, games, face painting, and a magic show.

The next CRN Family Conference will be held in downtown Chicago, IL in July 2015.



Children enjoying many wonderful daycare activities. Thank you! Laurie Smith and your amazing team.



Kenadee Julian and Maggie Haynes



Caroline Larimore



Lesli King from Sigma Tau

Thanks for the Memories:
2013 CRN Family Conference
"Heroes Among Us"
Washington, D. C., July 18th-20th



Batman with Mason and Livia Stilke and Victor and



Colleen Hammond, Carol Hughes, Pam Woodward, holding Sookie Boyer



Dr. Bill Gahl speaking on Friday

Welcome Reception & Family Introductions



Elva Smith



Batman



Dr. Jess Thoene and Jen Wyman



Rachel DaLomba and daughter Maya



Karen, Lily and Eric Schleuder



Garrett Thomas



Anne Stinson and Dr. Bill Gahl



Jose' Morales, Mick Swift from Cystinosis Ireland and Christy Greeley



Laurie Smith , Jeff and Jessica Larimore



Peytan Taylor



Victor Gardner and Aiden Smith

Making new friends,
reconnecting with old ones,
finding hope.



Aron Bjornsson from Iceland



Bryan Stout, Shannon Keizer, Brianna Smythe,
and Jennifer Loglisci



Conner Mills, Kim Forrester and Sarah Forrester

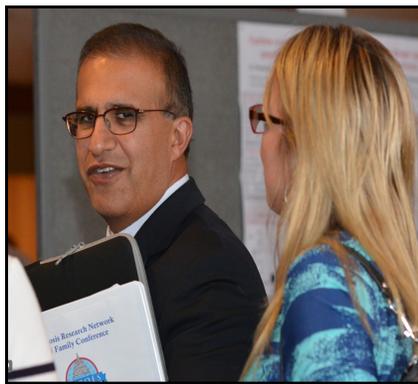


Maya Doyle and Steve Schleuder

Knowledge and Treatment Empower Us All, Thank You!!



Dr. Rick Kaskel and Dr. Jess Thoene



Dr. Ranjan Dohil



Dr. Richard Simon



Medical panel answers questions from attendees



Stephanie Cherqui, Ph.D.



Dr. Doris Trauner



Cystinosis Adult Panel answer questions from attendees



Joan and Anthony DeCroce



Sarah Larimore, Taylee Julian, Brooke Smith and Lily Haynes



Joy Bryant, R.N. and Dr. Galina Nesterova enjoy the moment.

Daycare
Dinner and
Dancing!
Fun for all ages



Martina Sigler



The Dinner /Dance on Saturday night was enjoyed by all!



Sookie Boyer with mom, Tahníe Woodward.

Research Update

by **Christy Greeley, Executive Director and Vice President, Research**

The Cystinosis Research Network's vision is the discovery of improved treatments and ultimately a cure for cystinosis. CRN utilizes a Scientific Review Board, comprised of leading experts on the disease, which reviews grant proposals and submits funding recommendations to CRN. More specifically, the SRB provides independent, objective review and recommendations regarding each research proposal, utilizing grant review guidelines established by CRN in accordance with the mission of the organization. The SRB also provides strategic advice and direction for the organization in terms of research topics to pursue.



Alex, Christy, and Jack Greeley

A major focus of CRN continues to be a determined effort to secure a promising future for the cystinosis community through the support and funding of research grants, fellowships, and other research based activities that lead to improved treatments, better quality of life, and ultimately a cure for cystinosis. CRN has funded close to \$4 million total in research grants and fellowships. Information on all current and past funded projects can be found on the CRN website under the Research tab, including grant guidelines, updates, and study recruitment information. Current grants funded in 2013 total \$270,000 and include:

- **Extension of Genotype-Phenotype in Egyptian Patients with Nephropathic Cystinosis.** Neveen Soliman, MD and Elena Levtchenko, MD, PhD, Center of Pediatric Nephrology and Transplantation, Cairo University, Egypt and Department of Pediatric Nephrology University Hospitals, Leuven, Belgium. Total Award: \$4,400
- **Extension of Proteomic Investigation of Cystinotic Cells and the Effects of Cysteamine Treatment.** Jill Jobson, Noel Carter, Achim Treumann, Ken McGarry and Rosaleen J. Anderson, Sunderland Pharmacy School, University of Sunderland. Total Award: \$55,000
- **Extension of the Continuation of Feasibility of Cystinosis Replacement Therapy in Cystinosis.** Jess G. Thoene, MD, Director, Biochemical Genetics laboratory, Active Profession Emeritus of Pediatrics, University of Michigan. Total Award: \$81,000
- **National Institutes of Health Cystinosis Fellowship, 2013 – 2015,** Galina Nesterova, MD, Mentor, William A. Gahl, MD, PhD. Total Award: \$130,000

As you can see, CRN has funded a variety of clinical and basic research under investigators all over the world. This work has led to many publications, which can be found on the CRN website under the Research tab in the Article Library, which also houses the majority of the most important cystinosis publications, including the recent clinical and parent guides to cystinosis care published by CRN.

Another guide is in the works to accompany the existing Cystinosis Parent Handbook, Cystinosis Standards of Care, and Cystinosis Transitioning Guide for Patients and Families. A workgroup is finalizing a Transitioning Guide for Physicians which will be a handy, one page reference which will be distributed to pediatric and adult care physicians involved in the transfer of care from the pediatric to adult care worlds.

These guides are an invaluable resource which we have distributed widely. We are fortunate to have the advice and experience of our Scientific Review Board, Medical Advisory Committee, and Professional Advisory Committees who are so committed to our community and have generously donated their time and expertise to ensure that this information is well documented and available to families and physicians.

Finally, I would encourage all of you to utilize the expertise of our CRN Cystinosis Fellow at the National Institutes of Health, Dr. Galina Nesterova. She has been working closely with Dr. Gahl over the years, treating the cystinosis patients seen under the NIH protocol, participating in CRN Family Conferences, and speaking at professional meetings on cystinosis. Dr. Nesterova has become an expert in cystinosis and is available to consult with any family or health care provider who needs assistance in the diagnosis and treatment of cystinosis. She can be contacted at nesterovag@mail.nih.gov. Please feel free to contact her with any questions or concerns.

Krummenacker, Sigma Tau (continued from page 7)

On a personal note, my daughter Laura was one of the original patients in the protocol at the National Eye Institute at the NIH. To be able to stand in front of a room filled with over 400 people and publically say thank you to Sigma Tau on behalf of all of the families was truly an honor. What made the evening even more special was to be on a stage with Dr. Jess Thoene, who received an award for his Vision on Behalf of Patients. He spoke about his admiration for the patients, the families and care-givers in the Cystinosis community and all of the work that continues for families. The evening was once again..... extraordinary.

NORD continues to work closely with both government and industry in all aspects of the rare disease community. The times we are in now with the implementation of the Affordable Care Act and all the fall-out that stems from beginning anything new, makes the role of NORD even more critical and to have a voice in Washington. NORD continues to be that well respected voice in not only Washington but around the world. Again it is an honor to be able to represent the Cystinosis community on the Board of Directors of NORD.



Gianfranco Fornasini from Sigma Tau, Jeff Larimore, Marybeth Krummenacker, Christy Greeley, Dr. Bill Gahl, Dr. Jess Thoene, his wife, Mari-jim, and Jose' Morales, enjoy NORD's 30th Anniversary celebration. In Washington, D. C.

KEEPING TRACK OF CYSTINE



A special blood test can give you important information

Cystinosis is a challenging condition that can cause serious damage to your body.^{1,2} One of the ways to prevent the damage is to keep your cystine levels as low as possible.³ But how do you know what your cystine levels are? For that, you need to have a special white blood cell (WBC) cystine test.

Why should I get tested?

In cystinosis, toxic levels of cystine can build up throughout the body.¹ If you delay taking your cystinosis medicine for even short periods of time, the cystine can build up.¹ The WBC cystine test tells you what your cystine levels are by measuring how much cystine has built up in your white blood cells.⁴ Getting regular WBC cystine tests can help you stay on top of how your cystinosis medicine is working and how your body is responding to it.⁵

Learn more about
WBC cystine testing
and other information
about cystinosis.

Visit KnowCystinosis.com

People who don't have cystinosis normally have cystine levels of about 0.2 nmol $\frac{1}{2}$ cystine/mg protein.⁶ People with cystinosis can have up to 100 times the normal amount.⁷ Many doctors say it's a good idea to get your cystine levels to less than 1 nmol $\frac{1}{2}$ cystine/mg protein.⁷

When should I have the test?

It's important to have the WBC cystine test done at the right time—based on your doctor's advice.

Your doctor may recommend having a WBC cystine test at least every 3 to 4 months.⁸ That way, results can be compared to get a clearer picture of how your medicines are working over time. Your doctor can talk with you about what your levels should be and may adjust your medicines as needed.⁴

How is the WBC cystine test done?

A sample of your blood is collected and prepared with a specific process to ensure accurate results. Then the blood sample is sent to another laboratory to be analyzed. When the results are ready, your doctor will discuss them with you.



At your next appointment, ask your doctor about WBC cystine tests and how often you should have them. You can work together to help you stay healthy—even with cystinosis.

References: 1. Levchenko EN, van Dael CM, de Graaf-Hess AC, et al. Strict cysteamine dose regimen is required to prevent nocturnal cystine accumulation in cystinosis. *Pediatr Nephrol.* 2006;21:110-113. 2. Nesterova G, Gahl WA. Cystinosis: the evolution of a treatable disease. *Pediatr Nephrol.* 2013;28(1):51-59. 3. Gahl WA, Thoene JG, Schneider JA. Cystinosis. *N Engl J Med.* 2002;347(2):111-121. 4. Dalton N. The importance of accurate cystine level testing. https://cystinosis.org/images/research/article-library/cystagon/2009_01_Cystine_Level_Testing.pdf. Published January 2009. Accessed September 9, 2013. 5. What are blood tests? National Heart, Lung, and Blood Institute. http://www.nhlbi.nih.gov/health/dci/Diseases/bdt/bdt_what.html. Published January 6, 2012. Accessed August 22, 2013. 6. Goodyer P. The history of cystinosis: lessons for clinical management. *Int J Nephrol.* 2011;2011:929456. doi:10.4061/2011/929456. 7. Bellidina EB, Huang MY, Schneider JA, Brundage RC, Tracy TS. Steady-state pharmacokinetics and pharmacodynamics of cysteamine bitartrate in paediatric nephropathic cystinosis patients. *Br J Clin Pharmacol.* 2003;56:520-525. 8. Procysbi [package insert]. Novato, CA: Raptor Pharmaceuticals Inc; 2013.

Hinsdale South Twins Celebrate Sweet 16 with Cystinosis Fundraiser

by **Sandy Illian Bosch** | sbosch@pioneerlocal.com | [@sandydoings](https://www.instagram.com/sandydoings)

Twins Anjie and Nelly Polanco, with little sister Maya, have decided to celebrate their 16th birthdays with a fundraiser rather than a party. The Hinsdale South sophomores have cystinosis and hope the money they raise will help in the search for a cure. | Sandy Illian Bosch/Sun-Times Media

Hinsdale South sophomores are thinking about what they can give.

Anjie and Nelly received their greatest gift nearly six years ago when each girl received a life saving kidney from the same donor. Their lives changed that day, but what didn't change was their diagnosis. The identical twin sisters have cystinosis, a genetic disease that causes crystals to form in the body's vital organs.

Anjie and Nelly decided together to forego the usual sweet 16 bash for a simple fundraiser to help end cystinosis. They launched a web page and took to social media to spread the word to family and friends.

"I've had five shares already," Anjie said of her Facebook post about the fundraiser.

Both girls have set a goal of \$1,600, all of which will go to the **Cystinosis Research Network** (CRN).

Advances in treatment of cystinosis have given the girls a couple of gifts already this year. New eye drops dissolve crystals that form in the eyes and cause vision problems for cystinosis patients. In just a few weeks the girls hope to start on a new drug that promises to fight the symptoms of their disease without the side effects, such as nausea.

Their medication must be taken every six hours, day and night. The new medicine can be taken just twice a day, promising the girls their first full night's sleep.

"There is just so much to celebrate and be thankful for this year and it is only natural for a me to want to give them the ultimate sweet 16 party," the twins' mom, Rachel DaLomba, said.

But she is not surprised by her daughters' decision.

"Anjie and Nelly are best known for their selflessness," she said.

Whether the \$3,200 is raised by the girls' Nov. 10 birthday, DaLomba said they likely will extend the fundraiser to their next birthday — Dec. 5. That's the day they received the kidneys that have allowed them to become sweet 16.



Twins Anji and Nelly with little sister Maya hold a Birthday Fundraiser for CRN



Anji and Nelly Polanco both have Cystinosis.

FDA-approved Cysteamine Eye Drops Now Available

Press Release

CYSTARAN™ (cysteamine ophthalmic solution) 0.44% is available in the U.S. for the treatment of corneal crystals in patients with cystinosis.

Corneal cystine crystal accumulation is a common symptom of each of the three forms of cystinosis – nephropathic, intermediate and nonnephropathic.

The formation of cystine crystals within the corneas of the eyes may lead to photophobia, or sensitivity to light. Other effects include eye pain, damage to the corneas, foreign body sensation (the feeling of something in the eye), and squinting. These may be debilitating to the patients who are affected by the crystals.

CYSTARAN is the first and only FDA-approved therapy for the treatment of corneal cystine crystal accumulation and is available to cystinosis patients in the U.S. with a valid prescription.

The most frequently reported ocular adverse reactions occurring in $\geq 10\%$ of patients were sensitivity to light, redness, eye pain/irritation, headache, and visual field defects.

Consult with your treating physician to see if Cystaran is appropriate for you.

Patients, caregivers and physicians in the U.S. can call the CYSTARAN Hotline (1-800-440-0473) through Accredo Specialty Pharmacy. The hotline is staffed with customer care representatives, pharmacists and nurses to process, counsel and answer any questions you may have about CYSTARAN from 8 AM-5 PM Central Time.

To enroll in the Cystaran program, you must have a valid prescription for Cystaran from your treating physician who will need to fill out the enrollment form and fax it to the number on the bottom of the form. The enrollment form is available at www.accredo.com in the physician section. Upon receipt at Accredo, representatives will research benefits for CYSTARAN, explain any financial responsibility patients may have and directly refer patients and caregivers to assistance programs available for copays and for those patients who are uninsured or underinsured.

Fulfillment and delivery of CYSTARAN will be direct to the patient. Due to the temperature sensitivity of CYSTARAN, it is shipped in a custom shipping container allowing for dry ice to keep the CYSTARAN frozen for up to three (3) days. In the box will be a temperature monitor that will be able to tell you if the product went out of the defined temperature range during the shipping process. This allows the product to be shipped all over the U.S. and stay within the specified storage temperatures.

CYSTARAN bottles should be stored, unopened, in the freezer. When ready for use, thaw 1 bottle for approximately 24 hours (do not heat the bottle or its contents). Thawed CYSTARAN can be stored between 36°-77° F for up to one week. Once a bottle has been opened the CYSTARAN solution is only stable for 1 week and the bottle should



Ask your doctor about
CYSTARAN

The ONLY FDA-approved
ophthalmic therapy for corneal
crystals in patients with cystinosis.¹

[Learn More](#)

be thrown away after being open for 7 days no matter how much medication is used. There is a place on the bottle to write the discard date as a reminder.

CYSTARAN should be given as 1 drop in each eye every waking hour, unless your doctor gives you other directions. If you wear contact lenses, you should remove you contacts before using CYSTARAN. You can reinsert your contact lenses 15 minutes after administration of CYSTARAN.

You will receive a reminder call from a specialist at Accredo when you are due for a refill, but are always welcome to call the hotline with any questions.

Sigma-Tau Pharmaceuticals remains dedicated to cystinosis patients, healthcare providers and the community through continued communication, education, support and access.

To obtain more information about CYSTARAN please call Accredo Specialty Pharmacy: 1-800-440-0473.

To contact Sigma-Tau, please contact Lesli King, Senior Manager, Patient Affairs: 1-301-670-5450 or Lesli.King@sigmatau.com.

Please see full prescribing information at www.cystaran.com

2013 American Society of Nephrology Conference in Atlanta

by Terri Schleuder

This year marked my first at an American Society of Nephrology (ASN) conference representing CRN. I was joined by veterans, Pam Woodward and Frankie McGinnis. For me, it was another "stepping out of the box" experience, but early anxious nerves soon turned into excitement and a feeling of empowerment; the opportunity to make a difference.

Over the course of three days of exhibition we handed out dozens of copies of the Standards of Care in Cystinosis, our Parent Handbook, the Transition Guide, Brochures, and Newsletters to physicians from around the world. A physician from India relayed there are 16 patients he treats and asked for something small to give them. He went home with 16 green puzzle shaped CRN key rings. A doctor from Russia relayed how difficult it is getting medication for his patients. We gave him all the resource materials we had. Another physician started to quickly walk by our booth murmuring cystinosis was just a pediatric disease. We quickly informed her over the last 25 years it is now and adult disease as well. She stopped, listened and took our materials with her.

One by one this exhibition experience continues to educate our adult nephrology physicians to the needs of our growing cystinosis adult population. For me it was also a time to reconnect with dear friends who have become "family" on this lifelong journey we all share. I left Atlanta with wonderful memories and a sense of accomplishment. It felt great!



Pam Woodward, Frankie McGinnis, and Terri Schleuder at the 2013 ASN conference in Atlanta, Georgia

"Costume Crazies" 5K Fun Run/Walk in Honor of Mason Reed, Held April 27, 2013

by Deb Reed

"Old Route 66, The Mother of the US", was sporting a new look on April 27th, 2013 in Vega, Texas. Vega is 13 miles east of the "Midpoint of Route 66". Route 66 stretches from Chicago, Illinois, to Los Angeles, California. Forty one participants filled the streets to participate in the first ever "Costumes Crazies 5 K Fun Run/Walk. Mason Reed was the honoree of the occasion and 22 members of his family all worked together to make the day a success.

The race began at 10:00 am with participants from surrounding towns in the Texas Panhandle. Friends showed up in crazy costumes, hats, socks, tutus, and camouflage suits to help us raise donations for Cystinosis, by walking, jogging and running in the race. They were entertained along the route at each turn with a Santa, a Funny Crazy Lady, Gypsies and Fire trucks. There was a water station at the half way point and fun handouts along the way. Love to Run Bracelets, bows, bells and leis with "Thank you messages for supporting Mason and Cystinosis!"

Medals for 1st, 2nd, and 3rd place were presented. Our 1st place winner was a 12 year old Vega Longhorn, Rhett Blasingame, with a fantastic time of 22.10 minutes.

We had plenty of help from the community and a special thanks to the Oldham County Chamber of Commerce for Co-sponsoring our Fun Fun. The run ended with a drawing for donated items and was a huge success. We raised \$4,402 for CRN. We look forward to our 2014 "Costume Crazies" 5-K next spring.



Deb Reed with Friends and Family at The "Costume Crazies" 5K Fun Run/Walk



Ryan and Kylee Reed with sons, Jaxon and Mason

Mason Reed Donates \$20,000 to CRN at Family Conference

by Kylee Reed

(Note: There was not a dry eye to be found during Mason's beautiful and heartfelt video and check presentation of \$20,000 to CRN at the Family Conference.)

Mason Reed began his stock show career when he was eight years old. He has had great success with many ribbons and buckles to show for it. Mason has a passion for the show ring that is undeniable.

(Continued on page 24)



Mason and his award winning pig in San Antonio.



Mason Reed presenting a check to CRN for \$20,000!

Cystinosis Standards of Care

The Cystinosis Standards of Care were written to help individuals with infantile Nephropathic cystinosis, their families, and their medical team. The information presented here is intended to add to conversations with physicians and other health care providers. No document can replace individual interactions and advice with respect to treatment.

One of our primary goals is to give affected individuals and their families greater confidence in the future. With early diagnosis and appropriate treatment, there is more hope today for families with cystinosis than ever before. Research has led to better methods of diagnosis and treatment. Knowledge is increasing rapidly by virtue of the open sharing of information throughout the world among families, health professionals, and the research community.

We acknowledge the important contributions to the Standards of Care of Dr. Galina Nesterova and Dr. William Gahl of the National Institutes of Health, and the members of the Cystinosis Research Network's Medical and Scientific Review Boards.

Cystinosis Standards of Care are available on the Cystinosis Research Network Website at:

<https://cystinosis.org/images/what-is/Cystinosis%20Standards%20of%20Care%20June%2019%202012.pdf>

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 the CRN website via:
<http://bit.ly/15SSH2K>

Checks payable to CRN—Keith and Billy Croce Memorial

Mason Reed (continued from page 22)

Last spring he made the Texas show circuit including visits to Fort Worth, San Angelo, San Antonio, and Houston. He also showed at local jackpots and his county show. When Mason began his career he said that if he made it in the sale at a major stock show he wanted to donate half of his proceeds to the CRN. His success in the ring reaped great reward at the San Antonio Stock Show and Rodeo. His pig, which was of the Yorkshire breed, placed 5th among some of the best in the nation. Going through the premium auction was an awesome experience for the whole family. Mason has now achieved one of his goals and hopes that his success will continue in the future.



Mason Reed and family present a check for \$20,000 to CRN's president Jeff Larimore.



Mason's generous gift to CRN

BMW Championship PGA Golf Tournament Volunteers Raise Nearly \$2,000 for CRN

Volunteers worked the concession stands during the BMW Championship PGA Golf Event, held in September at Conway Farms in Lake Forest, Illinois on behalf of the Cystinosis Research Network. As evidenced by these photographs, it was a fun way to support CRN. Tips and a percentage of the concession proceeds based on the number of shifts worked by volunteers raised nearly \$2,000 for CRN. We are appreciative of The Village Club of Lincolnshire's efforts to assist volunteer recruitment for this event.



BMW Volunteers have fun and help to raise almost \$2,000 for CRN.



7th ANNUAL-TEAM KACY-CYSTINOSIS 5K FUN RUN



Kacy Wyman and friends enjoying a beautiful day in southeast Michigan and raising over \$32,000 for Cystinosis Research Network at their 7th Annual 5K Fun Run!

by Jen Wyman

Sunny skies shone over Bloomfield Hills, Michigan for the 7th annual Team Kacy Cystinosis 5K Fun Run honoring Kacy Wyman (age 11). Over 400 smiling faces came out on that beautiful May day to support our family and the Cystinosis Research Network. Many were friends and family who have never missed a run. It has become somewhat of a tradition for people in our community. They set aside their morning on the first Sunday in May to help us celebrate Kacy's life and her battle with Cystinosis. It's a special day for all of us. This year was our biggest year..both in numbers of participants and in donations. We raised over \$32,000 of which every penny goes to CRN. This organization has been our lifeline since Kacy's diagnosis and it is an honor to be able to support and give back to something that has been such a gift to our family and to Kacy's health.

Next run: Sunday, May 4, 2014



CRN Cystinosis Awareness Bracelet Update

by Terri Schleuder

Thank you! Over the past year over 100 bracelets have been sold raising over \$1,000 for CRN. Your support has been amazing. We will keep a few bracelets on hand if anyone else is interested in purchasing one for \$20 each with \$10 of each purchase going to support CRN. Please contact me at: tschleuder@cystinosis.org for information. Be sure to put CRN Bracelets in the subject line.

Financial Update

by Jenni Sexstone—Treasurer

For the 9 months ended September 30, 2013



Jenni Sexstone and her sister Heather Vest, at CRN Family Conference.

We are pleased to present the financial results of the Cystinosis Research Network through September 30, 2013. 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 ended September 30, 2013, total income secured of \$368,000 and was approximately 38% more than the same period in 2012. The increase is driven by an increase of \$70,000 in grants received in support of the biennial family conference from Raptor Pharmaceuticals and Sigma Tau Pharmaceuticals. Direct public support is currently showing \$40,000 of improvement compared to the same period last year related to increased sponsorship for the conference and increased private donations. Donations received from public organizations such as United Way, Network for Good and Missionfish were approximately \$5,000 which is a \$10,000 decline from 2012. Fundraising efforts have provided approximately \$193,000 in the first nine months, which is relatively flat to 2012 at \$195,000.

Education & Awareness expenses amounted to approximately \$203,000 through September 2013 compared to \$35,000 in the previous year, which is related to expense for the 2013 CRN Family Conference. General operating expenses for CRN through September 2013 of \$37,000 were approximately \$7,000 higher compared to the nine months ended September 2012 due to timing of accounting/audit services. Total expenses related to fundraising events are \$29,000 less than the first nine months in the prior year due to events occurring in 2012, but not in 2013.

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 \$134,000 have been made through September 2013 compared to payments through September 2012 at \$272,000. The institutions that have received research support from and will continue to receive research work from CRN in 2013 include:



Josie Sexstone

- VU University Medical Center, Leuven, Belgium
- National Institutes of Health
- University of Michigan
- University of Sunderland, Sunderland, Scotland

Throughout the newsletter will be more detailed information regarding the status and findings of each of the research programs in progress.

Among the continued 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 2013 and in the first quarter of 2014, \$189,000 of research grant commitments could be paid out. 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 groundbreaking research, it is imperative for all CRN members to ensure sustainability by remaining focused on continued and new fundraising and donation efforts.

	Jan. - Sept. 2013	Jan. -Sept. 2012
Total Income	\$ 368,288	\$ 267,752
Total Expense	\$ 381,554	\$ 373,516
Net Operating Income/(Loss)	\$ (13,266)	\$ (105,763)
Cash and Cash Equivalents		
Cash on Hand—Jan. 1	\$ 102,649	\$ 212,219
Net Change in Cash	\$ (22,483)	\$ (108,416)
Cash on Hand Sept. 30	\$ 80,157	\$ 103,803

Dancing the night away at the 2013 CRN Family Conference dinner dance.







**ANSWERING THE NEEDS OF
THE CYSTINOSIS COMMUNITY**

Raptor Therapeutics has introduced its patient support program, RaptorCares. RaptorCares offers cystinosis patients and caregivers the materials and resources they told us they wanted and needed.

RaptorCares is a program that relies heavily on the input of the cystinosis community. So enroll today.
IT'S FREE. IT'S QUICK. IT'S EASY.

TO ENROLL

 RaptorCares.com/QA2

 1.855.888.4004

At this time, RaptorCares is only available to people in the U.S. When you enroll, your information will be kept strictly confidential.

RaptorCares is compliant with all FDA laws and guidelines. Please see full privacy policy at RaptorCares.com/QA2

CRN Development Update

By José Morales—Vice President, Development

It is amazing how quickly time has passed since our last newsletter. The Development Committee continues to facilitate and coordinate our community's efforts in raising funds to execute CRN's stated Vision and Mission. It is important to recognize that a key differentiator for our organization is the breadth and depth of its stated Vision and Mission:

VISION - the discovery of a cure, development of improved treatments and enhancement of quality of life for those with cystinosis

MISSION - supporting and advocating research, providing family assistance and educating the public and medical communities about cystinosis

We would not be able to execute our programs and activities without strong support from all of you and our extended community. The following are the individuals providing the leadership for all of us in this area:

Vice President of Development - José Morales

Corporate Sponsorship - Jeff Larimore, John Maccarone, Deb Reed

Fund Raising - Katie Larimore, Shannon Keizer, Tim Miller, Briana Smythe, Jen Wyman

Administration - Christy Greeley, Jenni Sexstone, Jen Wyman

I would like to highlight the two new members to the Development Committee: Shannon Keizer and Briana Smythe. Shannon and Bri both reached out and volunteered to become involved with our Committee coming out of this year's Family Conference. Shannon is partnering with Jen Wyman and Bri partnering with Katie Larimore in leading our efforts with Fun Runs and Letter Campaigns respectively. The new ideas and energy they bring to the Committee has been nothing short of inspirational.

2013 has brought us a slew of highlights, you will read about individual events in this newsletter, but I wanted to share with you the fun we had at our Family Conference's Apple iPad raffle. Anjie & Nelly Polanco did a great job spearheading our "sales campaign". The suspense was high as we drew the winning ticket. Lawrence Chan won on the initial draw and graciously donated the iPad back to the organization for an additional drawing. "Muchismas gracias" Lawrence, you rock! The audience waited in suspense as the ticket was pulled out of the bowl and the number was read. There was loud applause as the winner stood up and walked to the front of the room to claim his prize. The stars had aligned that day and Garrett Thomas became the proud owner of the iPad. Folks, it does not get any better than this! **(continued on page 31)**



Dr. Kaskel with Velyna and Alex Morales



Jack Hammond and Jose' Morales



Steve Schleuder and Kacy Wyman at the 2013 Wyman Fun Run. Fun Runs are a great way to raise money for cystinosis research!

2013-2014 Fundraising Calendar

Date: 2013	Type	Event	Honoring	Sponsor
Dec. 7th	Multiple Events	Chandler's Chance ... A Christmas Palooza for Cystinosis	Chandler Moore	Clinton Moore
2014				
January	Multiple Events	Mason's Round Up a Cure for Cystinosis, Facebook Auction and Letter Campaign.	Mason Reed	Deb Reed
February 13th	Letter Campaign	Steven's Birthday Letter	Steven Schleuder	Terri Schleuder
March 8th	Dinner/Dance	Laura's Miracle at Milleridge Reception	Laura Krummenacker	Marybeth Krummenacker
March 17th	Letter Campaign	Jack's Birthday Letter	Jack Greeley	Christy Greeley
April	Multiple Events	Village Club of Lincolnshire	Jack Greeley	Christy Greeley
April	Fun Run	Costume Crazies 5K Fun Run/Walk	Mason Reed	Deb Reed
April 24th	Golf outing	"Swings for Sarah"	Sarah Larimore	Jeff & Katie Larimore
May 4th	Fun Run	Kacy's 5K Fun Run for Cystinosis	Kacy Wyman	Jen Wyman
May	Music Festival	MayHem! Music Festival	Sierra Ayers-Mutchler	Ayers & Mutchler family
June	Letter Campaign	Sarah's Birthday Letter	Sarah Larimore	Jeff & Katie Larimore
June 7th	Golf Outing	C.H. Robinson Cystinosis Charity Golf Tournament	Tahnje Woodward	Tim Miller
October	Multiple Events	Village Club of Lincolnshire Event	Jack Greeley	Christy Greeley
December	Dance	Chandler's Chance... A Christmas Palooza for Cystinosis	Chandler Moore	Clinton Moore

In addition to the specific events listed above, we have on-going fundraising programs:

1. Jordan's Cystinosis bracelets/Jewelry On-line Merchandise Campaign; Sponsor Barb Kulyk
2. T- Shirts On-line Merchandise Campaign; Sponsor Briana Smythe

Cystinosis Ireland—Fall 2013

by Sue McGuire

2013 marked our 10th Anniversary and we have taken the time to look back over our first decade to all we have achieved in that time.

Our biggest achievement has been contributing €1.2million to Cystinosis research, we are extremely proud of this. We are so lucky to have such loyal and generous friends and supporters who have contributed the majority of this money, which is supplemented by funding from the Irish Government's Health Research Board's grants also.

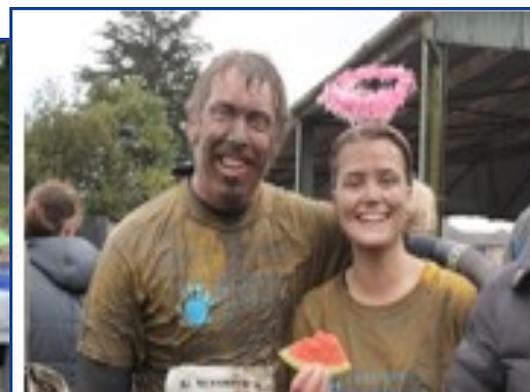


We looked back to when we hosted the fifth International Cystinosis Conference here in Dublin in June 2008, which was attended by over 200 delegates from many countries. We also reflected on the first Cystinosis Research Symposium which we held in the October 2011 with all our researchers contributing and reaching out to a new audience in University College Dublin.

We are also looking to the future and to funding more research and are particularly interested in more collaborative ventures such as that we undertook with CRN in 2011/2012. At this point too we would like to thank the Board and members of CRN for their on-going support and camaraderie.

But mostly we are reflecting on all the fun we have had during this ten years (along with the hard work!) and the wonderful people we have met who have given us their time, ideas, skills and money to help us continue to fund cystinosis research. The latest event we held in October was the "Runamuck Challenge" where Team Cystinosis comprised of 13 mad people who tortured themselves over a 5k (and 4 of them went round twice!) assault course which was difficult, cold, wet and MUCKY! However, they all enjoyed it and all were still smiling muddy smiles at the end! They also raised €3,687 for research (at today's exchange rate that's approximately \$4,970!).

Take a look!



Development Update (continued from page 29)

I am pleased to confirm the Committee has been successful in broadening the number of potential 2014 programs on the planning board that could be executed if we had events and families helping to raise funds for our organization. That said, we have a number of potential 2014 programs on the planning board that could be executed if we were able to increase the amount of funds raised this coming year. As such, we ask that you please consider becoming involved at whatever level you feel most comfortable with. The Development Committee is prepared to assist you in planning, executing and funding an event you are willing to sponsor. Please do not hesitate to reach out to me (E-Mail - jtm01@optonline.net or Mobile - 203 722-9292) to discuss how you can become involved.

We look forward to having the opportunity to work with you on becoming engaged and making a difference in the lives of our loved ones.



Team Cystinosis in Ireland raised almost \$5,000 for cystinosis research during their "Runamuck Challenge" fundraising event this past October.



Cystinosis Parent Handbook

The Cystinosis Parent Handbook is now available on the Cystinosis Research Network website at: <http://www.cystinosis.org/filemanager/Cystinosis-Parent-Handbook-Final.pdf>.

The Cystinosis Parent Handbook is a tool for families to use in their journey after receiving a diagnosis of cystinosis. Families are likely filled with questions about the disease and the future of their child's life. The Handbook will help families gain a better understanding of cystinosis and its treatment, learn to care for their child and find support from others in the cystinosis community. Families may want to keep the Handbook as a reference over the coming years.

The Cystinosis Parent Handbook was written with input from many members of the cystinosis community and the following contributors of the Parent Handbook Committee: Jean Blum, Maya Doyle LCSW, Christy Greeley, Colleen Hammond, Lauren Hartz, Jessica Britt Jondle, Marybeth Krummenacker, Katie Larimore, Mack Maxwell, Frankie McGinnis, Terri Schleuder, Steve Schleuder, Serena Scott, Sue Scott, Garrett Thomas, Lynn Thomas, Pam Woodward, Tahnne Woodward, and Jen Wyman.

Chandler Moore's Life Story

by Clinton Moore

Chandler was born on May 9th, 2005. He was a healthy, strong, lively boy weighing in at 10 lbs, 8 oz. As an infant I can remember him smiling and sometimes laughing out loud while he was sleeping. During the day we would take him outside under our big maple tree and he would stare up through it for what seemed like hours. I guess the way the wind would move the leaves just seemed to hypnotize him. He loved when we would yell as he would laugh when he heard the echo, and then ask us to do it again. One thing you could always count on from Chandler was a smile.

At 10 months old Chandler had to be admitted to the only children's hospital in Delaware, the A. I. Dupont Hospital for Children, 2 hours away. He had very excessive thirst and his lab work showed that something was wrong. Nine days later he was diagnosed with the a very rare disease called cystinosis. We were given the small textbook of instructions and sent home.

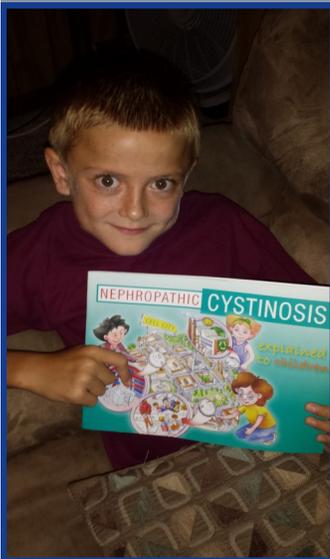
The next few months proved to be a little challenging. Getting into a routine and getting him to swallow those awful medicines seemed impossible. Sometimes we would squirt it in and he would spit it right back out. Eventually he got a g-tube placed for extra nutrition and it helped with the medicines.

Today Chandler is just as happy as he ever was. He loves to do most anything outside and I think his favorite activity is playing basketball or jumping on the trampoline. He is always running around with plenty of energy. Having cystinosis doesn't really seem to affect him much. I'm sure he would rather not take his pills or get his growth hormone shot every night but he just does it and moves on. Most people that meet him have no idea that he has this disease.

We did attend our first conference back in July. I think it meant a lot to Chandler to see other children taking their pills and getting their eye drops. Maybe a sense of not being the "only one". He made so many friends and actually cried when we had to leave. So many memories were made that weekend. We are already looking forward to the next one. Until then Chandler will continue to take pills and visit doctors. He will continue taking injections and doing eye drops. And you can be sure he's going to keep on smiling.



Chandler Moore



Chandler Moore, sharing his cystinosis story.



Chandler Moore enjoying a full and active life.

Why I am Thankful for my Disease

by Shannon Keizer

Now, you're probably thinking, "What? Is this girl crazy? Why would anyone be *thankful* for a disease?" Before I begin, I would like to clarify. There is a big difference between loving Cystinosis and being thankful for it. Although I do, on the occasion, jump out of bed and enthusiastically exclaim, "I LOVE the smell of Cystagon in the morning!!!!!" (cue sarcasm), I do not love the disease itself. I do not love the 'round the clock meds, gut wrenching side effects, frequent feeling of sickness, or any of the kidney issues. I am, however, grateful for the relationships, lessons learned, character building, and blessings bestowed in the complete package of Cystinosis.

We often find ourselves asking or even demanding "Why me? Why must I suffer like this? Why can't I be normal?" Lately, I've been contemplating these same questions with an entirely different mindset. "Why me?" has become, "Why am I the recipient of so many blessings? Why have I been given relatively good health when I was not expected to reach age ten? Why have I been granted this abnormally, extraordinary, full life?"

A common theme in all my speeches and writings about Cystinosis is this; embrace it. Embrace the good with the trouble. Embrace the entire package. I am reminded of the story of Job. After he was stripped of completely everything, aside from his own life, his wife questioned his integrity and ability to continue on. Job replied, "Shall we accept good (from God) and not trouble?" (Job 2:9-10).

It's true. Cystinosis drives me to do the unimaginable and has fueled my zest for life, love of adventure, and living to the fullest. However, far greater than any of these are the relationships and people that have been brought into my life through our common journey. I could continue trying to persuade you of the "good" of Cystinosis, but rather, I'd like to take a few minutes to *show* you.



Shannon Keizer and Katie Ahnen became friends because of Cystinosis.

Katie Ahnen and I met one year ago at a Cystinosis feedback session in Chicago. It took about 30 seconds to connect on a deep, profound level. Cystinosis has a way of doing that. Something so uniquely shared instantly forms a bond that most others can't imagine. With Katie, this was only the catalyst for greater friendship and connection. Throughout the course of the year we have become like sisters. The road between Milwaukee and West Michigan is well traveled. When communicating, rarely do we make mention of our mutual disorder. Instead, our discussions sound typical to that of most 20-somethings; school, work, boys, and stories of living out of our parents' basements. There are also those days when the phone rings, and all that's said is "I'm having a Cystinosis day." No other words are needed. One cannot deny the magnetism between those with Cystinosis. This past week, Jen Loglisci and I converged at Bryan Stout's crib in North Carolina. One day after arriving, I overheard Bryan's mom saying "I feel like I've know Jen and Shannon for ten years". This confirmed the instant sense of community and belonging. Our week of shenanigans included a Halloween party, Justin Moore concert, and a family trip to the South Carolina border, (just to say we were there). As the car overflowed with kids on laps and Bryan in the trunk, I apologized for having just taken my meds. Laughter and the aroma of Cystagon air freshener filled the vehicle. **(continued on page 34)**

Aside from the common med schedule, we rarely spoke of Cystinosis. It's like we are all brothers and sisters connected by a common gene. Actually, I think Bryan's dad tried to convince us we were all related. Although I'm not quite sure how that works...

Beyond Katie, Bryan, and Jen, the entire community and faces of Cystinosis have become my second family. If you have attended a conference, you may agree it is like a big family reunion. And how cool is it that our family reunions include the world's greatest doctors, researchers, and life changing presentations?



Jen Loglisci, Bryan Stout and Shannon Keizer visit in North Carolina.

This past July I attended my first conference solo. Each day a different "adopted parent" took me under their wing, something truly special. After that weekend, it just seemed natural to serve on the CRN development committee. I'm not gonna lie, my initial interest in joining was for the purpose of more frequent opportunities for communication and fellowship with an amazing team of people. As I am finding, when you combine passion, enjoyment, and a greater purpose, *that's* when magic happens.

So why am I telling you all this? Here's your take away; whether you are a caregiver or a Cystinotic, embrace it. Embrace the good with the trouble. Open yourself to new opportunities. Take a risk. Chase adventure.

This is my final charge. I challenge, no, I DARE you to reach out to someone in the Cystinosis Community. Go beyond the usual means of social media and pick up the phone, make a personal visit, attend a family gathering or conference. I guarantee you will walk away blessed and others will be blessed in return. My hope is that some day more people will be able to say, "I am thankful for my disease".



Young adults with cystinosis connecting at CRN Family conference in Washington, D.C. this past July. From left to right, Shannon Keizer, Briana Smythe, Steve Schleuder, Bryan Stout and Jen Loglisci.

Dr. Stephen C. Groft Honored with 1st Henri Termeer Lifetime Achievement Award

Press Release

The Global Genes | RARE Project (www.globalgenes.org), its Board of Directors and Honorary Advisory Board are proud to announce that Dr. Stephen C. Groft, Director, National Institutes of Health, Office of Rare Disease Research, will be honored as the first recipient of the Henri Termeer Lifetime Achievement Award for his nearly 30 years of service and commitment to advancing research and treatments for the millions of people afflicted with rare and genetic diseases. The award was presented to Dr. Groft at the 2nd Annual RARE Tribute to Champions of Hope™ Gala taking place at the Balboa Bay Club & Resort on Saturday, September 21, 2013, in Newport Beach, California.

“Dr. Groft is one of the original pioneers in the rare disease arena and is recognized worldwide as a leader in building collaborative relationships to improve patient treatment and care,” said Barbara Wuebbels, Associate Director Patient Advocacy and Investor Relations, BioMarin. “He has worked on both a national and global level towards increasing awareness and advancing research into orphan diseases and empowering patient advocacy organizations and patients alike. No one is more deserving of receiving this year’s Lifetime Achievement Award than Dr. Groft.”

At the 1st annual RARE Tribute to Champions of Hope™ event held in 2012, more than 400 guests were in attendance to celebrate the pioneering achievements of individuals and corporations in the rare and genetic disease community. The sold out event brought together rare disease stakeholders including patients, patient advocates, biotech and pharma executives, rare disease researchers, celebrities, professional athletes and philanthropists. Attendees were inspired by incredible stories of triumph and courage and were entertained throughout the night by special musical performances by leading recording artists including Chris Mann (Faircraft Records/Universal Republic), singer-songwriter Katrina Parker from NBC’s hit show “The Voice,” and recording artist and “American Idol” season 5 finalist, Elliott Yamin.

The 2nd annual RARE Tribute to Champions of Hope™ event will once again raise awareness for the nearly 7,000 different types of rare and genetic diseases that afflict roughly 30 million Americans and approximately 300 million people worldwide. The annual event is designed to bring together various stakeholders who Care About Rare™, while honoring many individuals from the rare disease community who are working tirelessly to advocate for the millions of patients and their families affected by rare and genetic diseases.

“The RARE Tribute to Champions of Hope™ Gala is a unique event that recognizes inspiring individuals who are working outside the box, innovating, and advocating for the millions of people impacted by rare and genetic diseases,” said Nicole Boice, President, Global Genes| RARE Project. “It’s a special time where we unite as a community and bring together new supporters and advocates to become part of this global movement.”

See link for video that features Marybeth Krummenacker. <https://cystinosis.org/news/media/136-cystinosis-and-crn-board-members-featured-in-dr-steve-groft-lifetime-achievement-video-at-global-genes-gala>



Dr. Stephen Groft, Director of the NIH's Office of Rare Disease Research.

Education and Awareness Update

by Terri Schleuder, VP of Education and Awareness

Since taking over the role of Vice President of Education and Awareness in July it has been quite busy. In August, after coming home from a very successful Family Conference, the education and awareness committee selected our 2013 CRN scholarship recipients. Elizabeth Patterson was awarded the scholarship for an individual with cystinosis and Kole Binger was awarded the Sierra Woodward Sibling scholarship. Read more about these fine young women on page four in this issue.



Terri Schleuder

On November 5th-10th CRN was represented at the ASN (American Society of Nephrology) medical conference in Atlanta, Georgia. This annual event provides an opportunity to educate and bring awareness about cystinosis to adult nephrologists concerning the needs of our growing adult cystinosis population.

Pam Woodward, Terri Schleuder, and Frankie McGinnis, manned CRN's booth answering questions, handing out Brochures, and recent publications including, the Standards of Care in Cystinosis, the Cystinosis Parent Handbook, and our Transition Guide. **(read more on page 21)**



Lily Schleuder

The end of the year was also busy collecting articles and pictures to format and prepare this newsletter. One new feature debuting in this issue is our Adult section. In it, we wish to highlight how our successful adults embrace life fully while managing cystinosis.

It is a pleasure to present two articles written by Shannon Keizer and Briana Smythe, two amazing young women who embrace life fully inspite of the challenges of living with cystinosis. I know you will enjoy reading their stories and insights.

We are also pleased to bring you Chandler Moore's story in our Family Feature section on page 32. We are always interested in sharing our families' journeys with cystinosis. They inspire us all. Please consider sharing yours in an upcoming issue.



Steve Schleuder

Owen's Outlook Holds 'Rock Out Cystinosis' Fundraiser

By Patty Blais

This has been a growing and developing year for Owen's Outlook. We, as a group, are still working towards obtaining our 501c3 status. This continues to be our ultimate goal as a group but we also wanted to begin our objective of raising awareness and initiative.

In May of this year, we kicked off our first official fundraiser, calling it Rock Out Cystinosis. Local bands donated their time and talent throughout the day. We also were fortunate to have a DJ/ Master of Ceremonies to fill in the down times between band set up and allow our guests opportunity to test out their singing talents with karaoke. Local restaurants donated enough food during the day for a buffet dinner and local pizza



Owen Blais with mom and dad, Nicholas Blais and Mariayh Barry.

places donated enough pizza to sustain our night crowd. The day was filled with raffles, guest appearances by local TV news personalities and Pepper Johnson, the lineman coach of the New England Patriots. Our turn out was amazing and a great time was had by all. This is definitely an event we want to grow and expand due to its overwhelming success. In late September of this year we had our first golf tournament. We had few sponsors due to our lack of non-profit status but none the less the event was a success. Monetarily we raised far more with our Rock out event, however, every golfer enjoyed their day and is looking forward to next year. They all offered ideas and future support for next year's event either through hole sponsorship or adding additional foursomes. There were 70 golfers at the event and an additional 15 people who came out to show support and share a meal. Due to that, we feel that this was also a success and



Musicians Rock it out for cystinosis.

a learning process.

Owen's Outlook has enjoyed much local support this year and our goal is to expand each of the events and look into other venues as well. It has been a very rewarding experience. We look forward to 2014 and the growing our venture in order to continue our support the Cystinosis community.



Family and Friends come together to support Owen's Outlook with 2 fundraisers for CRN.

September Chicago Area Family Dinner and Discussion



Dr. Langman describes the history of cystinosis to dinner guests.

A Chicago area dinner was held on September 17 at Maggiano's Restaurant in Skokie, Illinois, which included guest speaker and CRN Scientific Review Board member, Dr. Craig Langman. Nearly 50 people were in attendance, including 14 cystinosis families, nurses and physicians, and representatives from Raptor and Sigma Tau. CRN looks forward to coordinating patient gatherings like this in the coming months in other parts of the country.



Diana Hampton, Kristine David, Patrick Reichenberger, and Jenni Sextstone



Jeff Larimore with Rachel DaLomba and her twin daughters Anjie and Nelly Polanco



Kole and Dinny Binger, Eric Mosbrooke and guests.

Jack, Dave, Christy and Alex Greeley visit with guests at the Chicago area family dinner in September.





'Kickin' It for Jack Raises over \$6,000 for CRN



Jack and Christy Greeley at The Village Club's fundraiser, 'Kickin' It for Jack.



Jack Greeley and friends get ready to Kick it at the Village Club Fundraiser in his honor



Jack Greeley and Friends!

The Lincolnshire, Illinois community really kicked on Oct. 12 when over \$6000 was raised for CRN.

The Village Club hosted "Kickin' It For Jack" a kickball tournament fundraiser in honor of Jack Greeley. Some 150 players participated – including dozens of friends from Daniel Wright Junior High School, where Jack is an eighth grader. Plus, many more friends and neighbors came to have fun with food, drinks, raffles, a dj, and a balloon launch for Cystinosis awareness. Despite a rainy day, all stayed, played and supported Jack.

The amount of money raised would not have been possible with the sponsorship and support of many local businesses and organizations, including; Jelli Goods, Prairie House, Fresh Market, Wiener Take All, Sarpino's, Tamarak, Lincolnshire Sports Association, Lawrence Screw Products, The Prosk Family, Sam Martirano's, Eddie Merlot's, Bill's Pizza, DJ Ben Jamin, Robert Hancock, Jennifer Qualley, Laura Russo, Sue Wynne and many more individuals .

Even better, when asked to rate the day on a scale of 1 to 87, Jack gave it an 89.

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



Chandler Moore



Josei Sextstone and
Sarah Larimore



Dedicated to a Cure. Committed to our Community.

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

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.

Club Needed for Students with Chronic Health Disorders

(The article below was written by Christian Morales, a student at Occidental College who has cystinosis.)

by **Christian Morales**

September 24, 2013 Filed under **On campus, Opinions**



Christian Morales

At Occidental there is a strong focus on creating a culture in which students can thrive and learn from one another. Creating opportunities to see things through different lenses and gain new insights enables the Occidental community to become a stronger support system for individuals of all diverse backgrounds. Yet one area of diversity that seems to be missing or minimized at Occidental is raising awareness about individuals who have the additional challenge of managing chronic health disorders.

In addition to coping with all of the complexities and challenges of being a college student, individuals with chronic health disorders also have to be highly vigilant with everyday duties like monitoring vital signs, taking medications and scheduling medical appointments. There is also the pressure of being accepted for who you are, balanced with the fear of being ostracized for not fitting the profile of a stereotypical healthy college student. As a result, individuals with chronic health disorders struggle to create an identity that is independent from their disorder. The lack of student exposure and advocacy from the Emmons Student Wellness Center continues to exacerbate the problem. Yes, Emmons provides medical services to students such as "outpatient diagnosis and treatment of minor illnesses and injuries, clinical laboratory services and health education." But Occidental students lead none of these services, and they are poorly advertised. Students who have to deal with chronic health disorders on a daily basis have the potential to become targets for physical and mental harassment, and creation of a student-led club for those living with chronic health disorders would enable these students to organize group therapy sessions and casual get-togethers, and ultimately to feel comfortable and be more willing to share their obstacles and triumphs with the Occidental community. As an individual living with a chronic health disorder, I believe it is essential for Occidental to create a long-standing support system on campus. If Occidental did this, it would encourage all students to further explore, challenge and embrace differences at a heightened level.



Christian Morales with his dad, Jose'

Recent events confirm what is possible here at Occidental with regard to disease awareness. In the spring of 2010, the women's lacrosse team played the Purple Game in honor of Lauren Wemple '13. Lauren Wemple was recognized for her contributions as an athlete and for overcoming the challenges of having epilepsy.

In the fall of 2013, the women's soccer team partnered with the Global Genes Project, an organization that aspires to educate society about the multitude of chronic health disorders individuals live with to advocate and create a greater awareness amongst students.

The fact that these events took place presents a hopeful future but also highlights the need to create a student-run disease awareness organization on campus. The time has come for the creation of a club that will bring students who have chronic health disorders together to educate and increase the level of understanding. The creation of such an organization could allow the Occidental community and the student body to further support, understand and respect individuals that live with chronic health disorders. Such an organization would reflect Occidental's promise to enhance the student's education through "the fulfillment of individual aspirations and a deeply rooted commitment to the public good."



Christian on the far right with his sister, Alex, and parents, Velyna and Jose' Morales

Ignorance breeds contempt; people fear the unknown, and the unknown is anything but the norm. Perhaps the time has come for the Occidental community and administration to champion a campaign to enlighten all students. The creation of a student-run chronic health disorder support system would empower all individuals, not just those with chronic health disorders, to share their stories, enrich our school with awareness and explore the opportunities that Occidental has to offer.

Christian Morales is an undeclared sophomore. He can be reached at cmorales@oxy.edu or on Twitter at [@WklyCMorales](https://twitter.com/WklyCMorales).

- See more at: <http://occidentalweekly.com/opinions/2013/09/24/club-needed-for-students-with-chronic-health-disorders/#sthash.ftqZcoMr.tyZOpyAV.dpuf>

7th Annual C.H. Robinson Golf Outing Raises \$94,000 for CRN!!

by **Tahnie Woodward**

The 7th Annual CRN Golf Event was held at the Thanksgiving Point Golf Course in Lehi, Utah on Saturday, June 8th, 2013. Tim Miller and the Salt Lake Branch of C.H. Robinson spearheaded this remarkable day started in honor of Tahnie Woodward.

Jose Morales and Christy Greeley were in attendance this year, golfing and volunteering. It was great to have them there, showing appreciation for this wonderful fundraiser and all it does to help out the Cystinosis Research Network. The weather was gorgeous once again, with clear skies and no rain. A fabulous time was had by all! Many people put numerous hours of blood, sweat, and tears into this event. Most importantly, \$94,000 was raised for CRN! Check out this link to view a video of the event. <https://www.facebook.com/pokes#!/CHRobinsonFoundation>



Sookie Boyer and dad, Rory Boyer, enjoy the day.





C.H. ROBINSON

Presents the 7th Annual
**Cystinosis Charity Golf
Tournament**

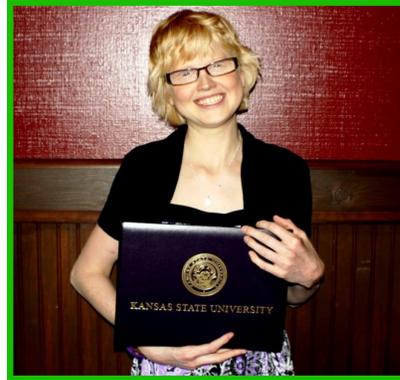
Thank you!
For your continued
and generous support



A great day for golf and fun in the sun at Thanksgiving Point Golf Course in Lehi, Utah on Saturday, June 8, 2013.

My Life Dealing with Cystinosis

by Briana Smythe



My name is Briana Smythe. I was diagnosed with Cystinosis/Fanconi syndrome when I was 15 months old. At first I seemed like a healthy baby girl. Then, my parents realized there was something wrong with me. I began to lose weight and not want to eat. All I wanted was water. I also stopped walking because of rickets.

Finally, after seeing many doctors and being flown to Kansas City, my parents were told I had cystinosis. This was not the diagnosis they were expecting; but at least their questions were finally answered.

My parents were told that I would need a new kidney by 10 years old, and would probably have to be on dialysis before then. My mom describes visiting the dialysis center with my older sister for a school field trip. After leaving the center, she felt sad. She describes it as a depressing environment. I thank God the doctor was wrong. Even though my parents weren't sure what my life would be like with this rare disease; they did what they had to do for me to grow up as normal as possible. I am now 26 years old and just now on dialysis waiting for my first kidney transplant. I have been on the list waiting for a kidney for a year now. I have been doing peritoneal dialysis since March, at home on my own schedule; which is great because of how busy I am. I don't like to let anything control my life!

"Determination"

As a child I didn't let things stop me from doing the same things as other children. I went to school. I played baseball/softball on teams with my friends. I rode my bike around my neighborhood, and even stayed over night at friends' houses once I was able to take my own medicines. I also tried out for basketball in middle school. I didn't make the team, but at least I tried. In high school I decided to get more involved in after school clubs to meet people. I joined the English club. We had to write poems and stories. This club actually helped me with the tough days of living with Cystinosis by writing my feelings out. My senior year of high school I went to New York with my English club. This trip was not only a big step for me; it was a big step for my parents letting me go without them. I had so much fun, I am so glad I didn't let my illness stop me from going.

"Support"

As a child I did have a lot of trouble with reading and spelling. From a young age I started working with a tutor. I also started helping with school plays behind the scenes,





Briana with her sisters and brother

and met some great friends.

I did have trouble in school because of the odor from the cystagon; and was bullied a lot because of it. In spite of this I was able to meet some great friends who didn't care about the odor, including my friend Catherine, who I met in Kindergarten. The funny thing is when we got older the doctor realized that she could not smell anything. We joke about that now, that no wonder she didn't mind my odor she couldn't smell it.

My family has also been a huge support for me. I have two older sisters and an older brother. My siblings have always been there for me. Even though my sisters live far away now and I don't get to see them very often, I know I can always talk to them. I know no matter what, we are family

and we love each other. I still think it's hard for my siblings and my parents to let me be independent because of my health problems. They are very protective of me. Sometimes I get frustrated because of this, but I try to remember they worry about me and my wellbeing. I am also really close to my aunt Sheila. She has always been there for me; even though she lives far away. It's great that I can still count on her if I ever need anything. I am so lucky to have an aunt who took me on a trip and made one of my childhood dreams come true. I had always wanted to swim with the dolphins. Thanks to my aunt that dream came true. I want to take a longer trip once I get my transplant. My aunt and I have talked about going to Vegas for a girl's only trip with my mom and sisters. Someday, I would love to go on a trip overseas or somewhere else just with my aunt. I really love my family, and any time I get to spend with them; I feel blessed.

"Hope"

I currently have two part-time jobs; Kohl's and Xcel, an after school mentor program. I've worked at Kohl's for two years and just love the people I work for. My manager tells us almost every day we are not only a team, we are family and we've got each other's back. It is so great to have a job like this. The mentor program I work for is also like a family to me. I volunteered with this organization while I was in college. Now, I am paid staff, but I would still work with the kids even if I didn't get paid. The director, Tina, is a big part of how I was able to go to the Cystinosis conference in DC. I told her I wanted to go to DC for this conference to meet others dealing with cystinosis. She set up an account for people to donate money for my trip. She then found a donor who matched whatever money I made from my fundraising. I don't know who that donor was, but I feel blessed that someone was so generous to do that for me.



Briana swimming with the dolphins

My Life (continued from page 47)

I feel lucky to have so many amazing people in my life behind me 100%! These people are what keep me positive even on my bad days. My Kohl's family was there when I got the call for a possible kidney match, and they were there when I found out I wasn't going to get the kidney this time. If it wasn't for them with their support and making me laugh about the situation I probably would have been upset longer. I realized that my transplant just was not meant to happen then. I may not have gotten that kidney, but I still feel I am lucky in many other ways. Some days my jobs are challenging, but I love what I do; and that's what matters.



Briana with her friend Catherine

"Admiration"

I'm also involved with the Big Brother/Big Sister program. This picture is of two amazing kids I met through the program years ago. I have known them since they were in grade school. I recently got involved again in the program, and have a new little "sister". I feel that it's important to volunteer to give back to others no matter what!



"Be Inspired"

I also decided to be on the Development Committee for the Cystinosis Research Network. My hope is that I am able to help educate people about this disease and raise money as well. I am currently working on getting fundraisers done with businesses in my town. I have one fundraiser for CRN on November 15th at Quiznos. I am hoping to be able to contact more businesses soon. So in between my jobs, volunteer work, people always ask me how I stay so positive. The truth is, some days I am not sure how I stay so positive, especially on the challenging days. I just keep reminding myself that there are others who are worse off than me. I also always smile because I want to show the kids I work with that no matter what their situation, you can still change your life and follow your dreams.



Briana with her nephew Brody.

I've spent many days in and out of the hospital with surgeries and other health issues, but I still made sure I did my homework so I could graduate high school with my peers, and finish college. It may have taken me longer to finish college because of health issues, but I never gave up! So last year I finally got my Bachelor of Science in Family Studies and Human Services!



Mitchell Smith with his dad Kent and brother Aiden Smith at the 2013 CRN Family Conference.



Taylee and Kenadee Julian with Jose' Morales at the 2013 CRN Family Conference

Cystinosis Research Network
 Family Conference 2015
 Chicago, Illinois
 (Details to follow in the upcoming months)



Dr. Craig Langman



The Croce Family



Mack Maxwell

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 _____

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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.

Agency Name	The Cystinosis Research Network, Inc.
Non-Profit Tax ID #	04-3323789
Address	302 Whytegate Ct., Lake Forest, IL 60045
Telephone	1-866-276-3669 (toll free), 1-847-735-0471
Fax	847-235-2773
E-mail Address	CRN@cystinosis.org
Web Page	www.cystinosis.org

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 Research Network
302 Whytegate Ct.
Lake Forest, IL 60045



Dedicated to a Cure. Committed to our Community.

Cystinosis is a rare, genetic, metabolic disease that causes an amino acid, cysteine, to accumulate in various organs of the body, including 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. The availability of cysteamine medical therapy has dramatically improved the natural history of cystinosis so that well treated cystinosis patients can live into adulthood.

CRN Vision and Mission

Vision. *The Cystinosis Research Network's vision is the acceleration of the discovery of a cure, development of improved treatments, and enhancement of quality of life for those with 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.*

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