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2013 CRN Family Conference Set for July 18-20 in Washington, D.C.



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

Please plan to join us to reconnect and 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 expert clinicians treating and researching cystinosis today.

More information on the Marriott Wardman Park can be found on their website at <http://www.marriott.com/hotels/travel/wasdt-washington-marriott-wardman-park/>

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



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



Dr. Galina Nesterova

NIH Clinical Expert, Dr. Galina Nesterova, Available to Cystinosis Community

Who do you contact when you have a question about cystinosis? There is help out there from your local clinician to facebook groups, but you may not think about one of the best resources available to the cystinosis community. Galina Nesterova, M.D. is currently being mentored by William Gahl, M.D. at the NIH and has become an expert in nephropathic cystinosis. She is committed to being a personal resource for families and individuals living with cystinosis. Those who have been under the care of Dr. Nesterova or have reached out to her find her to be knowledgeable and compassionate. Please don't hesitate to contact Dr. Nesterova at:

Galina Nesterova, MD
10 Center Dr. Building 10
Bethesda, MD 20892
(301) 496-9104
nesterovag@mail.nih.gov

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Message from the President

Christy Greeley

By Christy Greeley

2001 was a terrible year. In addition to the obvious tragedy of the 9/11 attacks that September, my son Jack was diagnosed with cystinosis in April, and he was very, very ill for two long years. That summer my husband and I decided to attend a cystinosis conference in Las Vegas at the suggestion of our geneticist. Jack was far from stable, but my mother-in-law bravely volunteered to stay with him and my 4 year old daughter Alex for a few days while Dave and I attended the meeting. It turns out that trip to Las Vegas changed our lives and connected our family with some of the most motivated, caring and brilliant people I've ever met. It also started my now 11 year journey as a board member for the Cystinosis Research Network.

CRN was in its infancy then, but concerned families banded together to develop the organization into what it is today. CRN had funded research for several years before that, but the new leadership worked to expand and formalize its efforts to provide greater family support and education and awareness programs. I agreed to serve as the first Vice President of Research and during that time helped to establish the first cystinosis patient advocacy group Scientific Review Board, headed by Dr. Bill Gahl, which has served as a model to other organizations in the manner it brings together experts in the field to review research proposals, ensure that a wide range of clinical and research issues are addressed, and encourages new investigators in the field of cystinosis. This is one of our achievements I am most proud of.

At that time, José Morales was elected first President of CRN. Due to his dedication and vision, CRN was able to achieve a higher level of advocacy. His leadership galvanized those families who knew what the community needed, but weren't entirely certain how to deliver it. He paved the way and has continued to provide invaluable counsel to me during my term as President. I am so grateful to him for everything.

So much growth has taken place over the last 11 years, all tied to CRN's mission to support research, family support and education and awareness activities equally. You've all heard the saying, "Knowledge is power". Conversely, sometimes "Ignorance is Bliss", but we all know that is certainly not the case when you are dealing with a diagnosis like cystinosis. CRN's goal has been to provide as many avenues as possible to provide that knowledge to individuals, families, professionals and the public. This has been accomplished over time due to the hard work of dozens of individuals with cystinosis, their families and a variety of professionals across the globe – here are some highlights:

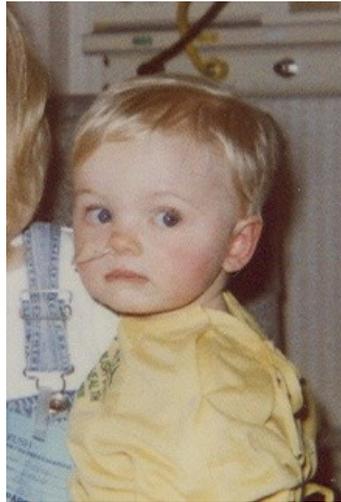
- A Board of Directors and Executive Committee were established which allows all cystinosis community members to serve and have a voice in the future of this disease. Board term limits were established, ensuring that a variety of individuals would be involved in an organization that serves as the voice of the community.
- Individuals with cystinosis have taken on high leadership positions within the organization, a key milestone to me which indicates that they are not just surviving into adulthood, but thriving. These individuals are vital, important members of our board of directors.
- CRN established a Scientific Review Board to review research proposals and Medical and Professional Advisory Boards to assist with the variety of other issues the community faces. These experts have been indispensable in the success of CRN, most notably with regards to their involvement in the Family Conferences. Their service is a gift to us all.
- The CRN website has been redesigned and updated continuously to ensure easy access to critical information. CRN has established a presence on Facebook and YouTube as well as valuable tools to disseminate information to a greater audience.

Continued on page 3

*denotes President Emeritus

Message from the President (cont.)

- The CRN Yahoo family support email group has been an active place for families to interact for many years.
- Six Family Conferences have taken place across the country since 2003, with a 7th scheduled for July 18-20, 2013 in Washington, D.C.
- A comprehensive contact information database of individuals with cystinosis across the world has been collected.
- Various cystinosis scientific symposia have taken place at the National Institutes of Health, in conjunction with family conferences, and internationally during medical society meetings, including recent CRN sponsored events at the 2011 International Pediatric Nephrology Association and the 2012 Latin American Pediatric Nephrology Association meetings.



Jack at diagnosis, April 2001



Jack at age 12, March 2012

- CRN has developed relationships with cystinosis advocacy groups from around the world, all of which are invited to attend and participate in our biennial Family Conferences. To this end, CRN has exhibited at cystinosis conferences in the Netherlands and in Ireland, will sponsor the Cystinosis Foundation Mexico's meeting scheduled for August 2012, and are also proud to have recently co-funded a research project by Dr. Besouw and Dr. Levtchenko with the Cystinosis Foundation Ireland.
- An annual Call for Research Proposals has been issued by CRN since 2003 which has resulted in the funding of millions of dollars in cystinosis research.
- An Article Library has been established on the CRN website which allows individuals to easily access a comprehensive collection of cystinosis research articles.
- The Pediatric to Adult Care Transition Guide was published in 2011, compiled by a CRN workgroup made up of families and individuals with cystinosis, medical professionals, and spearheaded by Professional Advisory Board member and social worker Maya Doyle.
- That same workgroup is currently working to produce comprehensive clinical care guidelines for all ages, to be published later this year. This is a groundbreaking project which will put vital clinical care information in the hands of families and caregivers.
- The Cystinosis Advocate, the CRN newsletter, is mailed twice yearly to over 5,000 individuals and past and present issues are available online.
- CRN representatives have exhibited at numerous professional meetings such as the American Society of Nephrology and the American Society of Pediatric Nephrology. The impact of a group of parents and individuals with cystinosis available to discuss cystinosis directly with doctors in the convention setting is unprecedented and incredibly effective for education and awareness of both pediatric and adult nephrologists.
- CRN has provided thousands of dollars in academic scholarships to individuals with cystinosis and their siblings.

Continued on page 4

Message from the President (cont.)

- In response to the need for a new generation of clinical experts in cystinosis, CRN established and funded the NIH Clinical Fellowship under the direction of Dr. Bill Gahl. Dr. Galina Nesterova serves in this capacity and has worked to not only become a clinical expert in the disorder, but has also spoken at countless grand rounds, medical conferences and CRN conferences, educating professionals and families. In addition, she has made herself available to consult with physicians around the world on cystinosis patient treatment issues. Dr. Nesterova has also been instrumental in the authorship of the Cystinosis Treatment Guidelines currently underway.
- CRN has become a leader within the rare disease community as a Member Organization of the National Organization of Rare Disorders and in our affiliation with the Genetic Alliance. CRN board member Marybeth Krummenacker currently serves on the NORD Board of Directors.

None of this would have been possible without massive fundraising by the community. You may have heard the term, "grassroots fundraiser", which is usually defined as a fundraising effort that derives most of its power and reason for being from a community. That is precisely how CRN is predominantly funded -- by the community, by families who have run countless golf outings, dinner dances, bake sales, hoedowns, family fun days, jewelry sales and 5K runs. Families have sent out birthday and holiday letters and hosted Facebook birthday fundraisers, asking friends and family to contribute in honor of their child. Eastside High School in Greenville, SC adopted CRN for their Wade Hampton Week and raised over \$207,000 – this fundraising provided a large portion of the funding for the NIH Clinical Fellowship. The C.H. Robinson Corporation in Utah has hosted a golf outing in honor of the Woodward family for years which has raised nearly \$200,000 for CRN. Family foundations, corporate matches and the United Way donor program have all provided significant funding towards our goals. Corporate partners, most notably Sigma Tau, Orphan Europe and Raptor, have also supported us generously, especially important in the funding of our family conferences.

The connections forged during this fundraising are powerful. And these connections we have developed over the past eleven years with individuals, families, organizations and professionals have enabled CRN to provide the services so vital to the community.

What does the future hold? New treatments are being tested, awaiting approval. Promising ideas for alternative treatments and even a possible cure are being tested. CRN will continue to support our families as we recognize that the daily needs and challenges we face are just as important as the research we fund. We will continue to expand our programs and services to reflect the ever changing needs of teens and adults with cystinosis as they thrive into adulthood and we learn more about this disease from these trailblazers.

I have been honored to serve on the CRN Board of Directors, especially these last five years as President and Executive Director. My greatest wish is that these accomplishments will be built on by my successor and that the cystinosis community will continue to grow and exhibit the strength of character I see each day. My final term ends this July, but I'm not going far -- I will serve as Executive Director during the transition as our new President acclimates to the position. What I will take away will be some of the greatest friendships a person could hope for, I am grateful to you all for sharing this journey with me and my family.

My warmest wishes to you all,

Christy Greeley

President and Executive Director

CRN Participates in the National Institutes of Health (NIH) Rare Disease Day 2012 Celebration and the Federal Drug Administration (FDA) – Rare Disease: Patient Advocacy Day

National Institutes of Health Rare Disease Day, Washington, D.C.

Christy Greeley and José Morales represented CRN during the 2012 Rare Disease Day activities. Rare Disease Day was established in 2008 with the objective of elevating public awareness about rare diseases, the challenges encountered by those individuals affected, the criticality and importance of research towards the development of diagnostics and treatments, and the impact these rare disorders have on patients and their families' lives. A large part of the success of Rare Disease Day is the collaboration and partnership of a wide array of organizations, companies and government agencies who share the goal of raising awareness for rare diseases.



On February 29, The National Institutes of Health, Office of Rare Disease Research sponsored the day-long symposium celebrating rare diseases research efforts across NIH, the Federal Government and the advocacy community. Dr. Steve Groft was one of the featured speakers and he quoted a familiar statistic in his welcoming remarks -- there are approximately 7,000 rare diseases, we know the molecular basis for about 4,000, and yet there are treatments for only 200 -- one of these is cystinosis. Also of note, Dr. Francis Collins announced the establishment of a Genetic Testing Registry, now available via the NIH website. More specifically, a wealth of cystinosis disease and testing information can be found there at <http://www.ncbi.nlm.nih.gov/gtr/diseases/C0010690/>

The **National Organization of Rare Disorders (NORD)**, one of the sponsors of the two day celebration, hosted a reception February 29th for patient advocate organizations and others participating in the NIH and FDA events. CRN was in attendance and continued our efforts to highlight and share information about CRN with other organizations and tangibly evidence our commitment to the broader rare disease community.

Federal Drug Administration – Rare Disease: Patient Advocacy Day, Bethesda, Maryland



Dr. Stephen Spielberg, Deputy Commissioner, Medical Products and Tobacco, delivers the keynote address as part of the FDA's Rare Disease Patient Advocacy Day

Patient groups who represent rare diseases are enormously important to the central mission of FDA in health care. Of the thirty new molecular entities that the FDA shepherded through regulatory review and approval last year, more than one-third were indicated for rare diseases. By better understanding this special community of patients, the FDA has further become better prepared to capitalize on novel technologies and emerging trends in personalized medicine, for the general treatment of disease.

On March 1, 2012, the FDA opened its door, under the banner of FDA–Rare Disease: Patient Advocacy Day, coordinated by the Office of Orphan Drug Products, to bring patients and their families together with FDA officials on the White Oak campus. FDA officials, along with experts from the patient advocacy community, provided a full day of lectures and round-table discussions that pertained directly to the needs of patients with rare diseases. The day was devoted to learning—about each other, about the strides being made on diverse fronts in health care, and about the ongoing need for better treatments and interactions with members of the rare disease community.

A highlight of the day for the CRN attendees was the keynote address given by Dr. Stephen Spielberg, Deputy Commissioner for Medical Products and Tobacco. Dr. Spielberg relayed a story about how the failure of a placebo controlled trial of Vitamin C in cystinosis, which he conducted with Dr. Schneider and Dr. Thoene in the 1970's, helped establish the first data monitoring board at the NIH. His point was that rare disease research often not only helps in the treatment of the particular rare disease, but also often expands more general medical knowledge -- and cystinosis certainly is a wonderful example of this. Clearly our cystinosis community is grateful for all of the talented individuals that have committed themselves to the study of cystinosis.

We were thrilled to participate in all three events and worked the entire time to ensure that CRN and cystinosis were visible to NORD, the NIH and FDA officials in attendance. Our continuing engagement, contributions and collaboration with the greater rare disease community are important towards accomplishing our mission of "supporting and advocating research, providing family assistance and educating the public and medical communities about cystinosis."

CRN Awards Scholarships to THREE Deserving Applicants!

Cystinosis Research Network is pleased to award scholarships to three individuals with cystinosis who are an inspiration. The three applicants each have unique strengths from each other and have all conquered the challenges of cystinosis to go on to post secondary schools. No one applicant stood out from the others, so CRN decided to award a \$1,000 scholarship to each of them.

Katie Ahnen



Katie Ahnen

Katie is a 2010 graduate of Arrowhead High School in Hartland, Wisconsin where she was captain of the Junior Varsity swim team. Katie is currently attending Waukesha Area Technical College where she has already become a Certified Nursing Assistant and is taking more classes with a goal of becoming a nurse. Katie works part-time in an assisted living home where she is passionate about helping the elderly.

Her academic advisor at WCTC stated, "Katie is a model student. She is always organized and willing to take on more than what is required of her...Katie is one of only a few students that have chosen to take on extra courses each semester that will benefit her in the nursing field."

Mikaela Gard



Mikaela Gard in the lab at Parkland College.

Mikaela is a 2011 graduate of Casey-Westfield High School in Casey, Illinois where she was a member of the National Honor Society, FCCLA (Family, Career, and Community Leaders of America), and Scholastic Bowl. She is attending Parkland College in Champaign, Illinois and is pursuing an Associates Degree as a Veterinarian Technician. Mikaela's dream is to work in a wildlife zoo or veterinary office to help animals in need, learn more about animals, and educate others about animals.

Mikaela's FCCLA teacher said Mikaela is one of the first to "step up" and always follows through on her commitments. Mikaela stated in her scholarship essay, "I do not mind having to fight cystinosis because I would rather fight cystinosis than let it beat me down."

Weston Tschannen

Weston is a 2011 graduate of Brookfield High School in Brookfield, Missouri where he was a member of the National Honor Society and active in golf, wrestling, and the culinary team. Weston is attending the University of Missouri-Columbia and majoring in Hospitality Management. Ever since he was nine years old, he has had a dream of running his own restaurant.

Weston received a kidney transplant from his mom, Barb, in December, 2009. Weston could no longer be a part of the wrestling team after his transplant, but after the team brought home a state championship, his coach asked him to come to the gym floor for their final applause of the season. His high school counselor stated, "Weston is an energetic, articulate, and responsible young man whose maturity, academic success, and positive attitude were evident not only to his peers, but also to teachers and support staff alike."

Congratulations to our Graduates!

High School Graduates

Chelsea Heath

Chelsea is graduating from Bastrop High School in Bastrop, TX on June 1. She plans to continue her education and is considering becoming a nurse or veterinarian.

Chelsea started showing symptoms of cystinosis at 8 months but was not diagnosed until she was 1 1/2 years old. She had a gtube and jtube most of her childhood and adolescent years. Chelsea had a kidney transplant in April, 2008 when she was 15.



Chelsea and her horse, Coco



Chelsea and her sister, Amanda

Melissa Hagen

Melissa graduated from Indian River Charter High School in Vero Beach, FL on May 19. She will be attending Indian River State College in Fort Pierce, FL to study phlebotomy and will continue on to be an Ultrasound Technician.

Melissa was diagnosed with cystinosis at 16 months old and was transplanted in April, 2008.



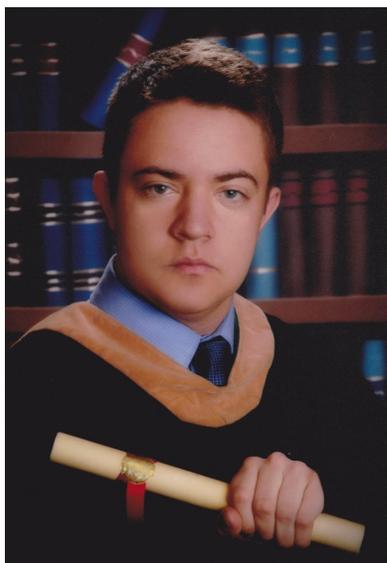
Melissa Hagen



College Graduates

Shea Hammond

Shea graduated from Merrimack College in North Andover, Massachusetts with a Bachelor of Science in Business and a minor in English. Shea was diagnosed with cystinosis when he was eight months old. He had a kidney transplant at age 16 and another at age 19.



Ashley Abedini

Ashley graduated from Wichita State University in Kansas with a degree in Marketing in May, 2012. Ashley was diagnosed with cystinosis when she was six months old. She had a kidney transplant on September 6, 2011 and has been doing great. One of Ashley's hobbies is skydiving.



Jessica Jondle Releases Book about Growing up with Cystinosis; proceeds go to CRN

Jessica's book has raised over \$5,000 for CRN so far! Order your copy today!

By Jessica Jondle

It was with great apprehension that I published my book, *Roller Skating with Rickets (and Other Paradoxes of Life with Genetic Disease)*, at the end of January. There were so many questions swirling around in my head after making it available: Will

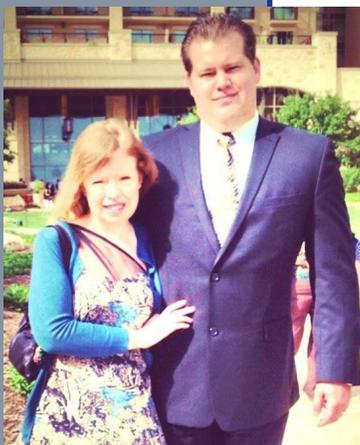


anyone read it? Is it any good? What if my students find out? By writing a book about living with cystinosis, have I made it such a monumental part of my identity that people won't understand that first and foremost, I am a junior high school teacher and dean, a wife, a friend, a workaholic, and an occasional adventurer? Will the book offer comfort to any of the mothers with young children with cystinosis, or is it presumptuous to believe that my life story could bring encouragement to anyone?

I have received answers to some, but not all, of these questions. I have been blessed in that many people have read RSWR. Last week, I learned that one of the parents at my school had discovered my book and blog online - and despite requesting that none of my students read it before either they graduate or I leave the school, I still worry that my life as "just the geeky history teacher" might hang precariously in the balance. I can't guess how others might see me (can we ever? should we even care?) and I still don't know if the book is any good, but I've started to realize that it doesn't matter. Let me share with you what I mean.

I was born at a military hospital owing to my father's career as a Naval officer. One of the difficulties of having all my young childhood health care handled at a military hospital was that the doctors there, as members of the military, would often serve short stints and then be stationed elsewhere. It was difficult to form connections with doctors who would remain in my life for a year, perhaps two or three. It is no surprise, then, that the doctor who diagnosed me when I was 22 months old was out of my life by the time I was four. And yet, God (and Google) allowed me to find this man a quarter of a century later. When I emailed him asking if he remembered me, his reply was immediate and positive: *of course*. I was able to send him a book, and after receiving his feedback, I know that this endeavor was meant to be and my doubts, meaningless. If Dr. Kerry had been the only person to ever read *Roller Skating with Rickets*, it would have been worth it. He wrote:

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Jessica and Wayne Jondle

Jessica Jondle Releases Book about Growing up with Cystinosis; proceeds go to CRN (cont.)

"One of the best rewards I have as a [now civilian] pediatrician is that I get to see babies grow up to be adults and with my now totally gray hair, I have seen a few. So I see myself often as an extension of their families. There is a connection of sorts and a respect (or love) that develops between us. The Army with its four-year assignments robs a pediatrician of that reward. I sadly did not get to see you grow up, but along came your book. You took me from very familiar ground, when you were young and through the diagnosis time of your life. I remember so much of that. Especially the conversation I had about allowing you to be active [roller skate] and dealing with any broken bones that may result. But you also took me into your life from that point on. In reading your book I could not help wondering what it would be like if we could just spend one day with God, watching Him weave the tapestry of events that we call life. Watching Him pick who we meet, when, where, and see the impact that encounter has on us years later. We all touch other people's lives. I happened to touch yours and you now are touching mine again. "In my 31 years of pediatrics, I have seen God's hand work many miracles and change many lives. I used to see bad things happen to good families. I would often find myself yelling at God about this as I felt it was so unfair. I finished reading your book at 3:00 AM this morning and as of right now I no longer see things that way. God has a purpose and there are reasons. I will no longer be an enabler but take on a more constructive role and become an encourager. I will even try not to use the phrase "special needs person" anymore. I will not limit God. We don't know it all, but He does.

"Your book helped rekindle a passion that was slipping from me. So this is what God gave me at 3:00 AM after reading your book: When we know that we can't even hold a tune, God gives us all a song to sing and yours is one of the sweetest I have heard. I hope someday I get to meet you and your husband, but if not, I will see you in heaven and we will take up where we left off. Love ya."

Cue the waterworks! It is without shame that I admit that I still cry when reading this incredible doctor's words, nearly three months after receiving them. I don't know if everyone will like my book. I can't know how many people will read it. But I do know, with this humbling note from Dr. Kerry, that writing it was *worth it*.

CRN Adult Care Excellence (ACE) Initiative

The Adult Care Excellence (ACE) Initiative continues to work to provide resources for the cystinosis community. Since the completion of the Pediatric to Adult Care Transition Guide (available on the CRN website), we have been developing a Guide for Parents of Children with Cystinosis and collaborating with Dr. Galina Nesterova and Dr. William Gahl at the NIH to develop Cystinosis Care Guidelines. The Guidelines are in a format that can be shared with physicians and other medical caregivers regarding the medical care of a person with cystinosis through all of life stages. The Cystinosis Care Guidelines will be available on the CRN website. We welcome new members to the committee and any feedback from the cystinosis community. The Guide for Parents of Children with Cystinosis should be available on the CRN website by the end of the summer. The chapters cover many subjects including:

- My child was just diagnosed. What do I do now?
- How to talk to others about cystinosis.. Informing family and friends
- Creating a care binder for your child
- Infancy and toddlers
- School - age
- Finding information and experts
- Family issues. Caring for yourself, your marriage, and your other children
- Templates for care binders, medication records, etc.
- Early childhood
- Early adolescence...and much more!

CRN Financial Update

By Jeff Larimore-Treasurer

We are pleased to present the financial results of the operating activities of the Cystinosis Research Network through February 29, 2012.

For the two months ended February 29, 2012, total income secured of \$21,548 was approximately 5% greater than the same period in 2011. The increase was derived from improved direct public support donations. Fundraising efforts from letter writing campaigns and sporting events accounted for 46% of the revenues received through February.

Total expenses incurred through February were \$13,348. Education and Awareness expenses for printing and mailing the Fall/Winter newsletter of \$8,363 was the material expenditure for this reporting period. CRN was also pleased to continue its scholarship program by awarding two \$1,000 scholarships. Operating expenses through February included Directors and Officers' insurance and annual organization registration fees.

Net operating income for the reporting period was approximately \$8,200. Net change in cash for the same period increased \$6,269 due to the operating activity referenced above.

As noted in the Fall/Winter newsletter, a challenge remains for the Cystinosis Research Network to provide payment for research grant commitments as they are progress billed. We estimate that approximately \$285,000 of grants payment requests will be made during 2012. In March and April, requests and payments were made to the University of Michigan and University of Sunderland of approximately \$89,000. In order for CRN to help those researchers and physicians who are focused on helping our children, adults and families, CRN's membership continually must be committed to sustaining its advocacy group through fundraising and donations.

In addition to fundraising and donations, efforts by the Executive Committee and Board Members will be extended to pursue public and private grants and to submit written requests for funding consideration to private foundations. The year end 2011 audit report and Form 990 tax return is available on the website for review. If there are any questions regarding those reports, please do not hesitate to contact me.

CRN Accepting Nominations for New Board Members



James Sausman, age 3

Have you ever thought about becoming more involved in your organization? Have you ever thought about becoming a member of the Cystinosis Research Network Board of Directors, helping to shape our group for the future? NOW IS THE TIME TO GET INVOLVED! CRN is accepting nominations for new board members. The work is not hard, the rewards are many. The board meets 3-4 times a year by telephone conference call. Once every two years the board has an in-person full board meeting that takes place in conjunction with the Family Conference. The Board of Directors will vote in new members in July, so think about it and let us know if you are interested.

CRN is looking for committed people who are willing to get involved and work to keep CRN the outstanding organization that it has become. Being a volunteer helps others, lifts your spirits and is really very rewarding. So please consider joining CRN, and let us know if you are interested.

"Service to others is the rest you pay for your room here on earth" —Mohammad Ali

CRN Exhibits at ASPN/PAS Meeting



Roisin Keon and CRN Board Member Jenni Sextone represented Cystinosis Research Network along with Colleen Hammond and MaryBeth Krummenacker (not pictured) at the American Society of Pediatric Nephrology (ASPN)/Pediatric Academic Societies (PAS) meeting in April, 2012. The annual meeting was held in Boston, MA.

CRN Exhibits at ASN Kidney Week



Dr. Rick Kaskel was one of many medical professionals who stopped by the Cystinosis Research Network booth at the 2011 American Society of Nephrology (ASN) Kidney Week Scientific Exposition in Philadelphia, Pennsylvania in November, 2011. Manning the booth were CRN Board Members Colleen Hammond, Marybeth Krummenacker, Karen Gledhill, and Pam Woodward (not pictured). ASN leads the fight against kidney disease by educating health professionals, sharing new knowledge, advancing research, and advocating the highest quality care for patients. Physicians who treat patients with cystinosis from more than 20 countries visited the booth.

CRN Research Update

By Elva Smith-Vice President, Research



Eddie Langley and his dog, Lestat

A major focus of the Cystinosis Research Network continues to be a determined effort to secure a promising future for the cystinosis community through the support and funding of research grants that lead to improved treatments and ultimately a cure for cystinosis. CRN has a current research commitment of over \$800,000.00 and has funded nearly \$3 million total in research grants and fellowships. Information regarding these research grants, progress reports, funding, and final reports is provided to the cystinosis community via our web site www.cystinosis.org.

Current Research Studies:

Neveen Soliman, MD and Elena Levtchenko, MD, PhD, Center of Pediatric Nephrology and Transplantation, Cairo University, Egypt and Department of Pediatric Nephrology University Hospitals, Leuven, Belgium, "*Genotype-Phenotype in Egyptian Patients with Nephropathic Cystinosis*", Grant awarded March 2012 for a 12 month period. Total award: \$15,750.

Martine Besouw, MD, Department of Women and Child Laboratory of Pediatrics, University Hospitals, Leuven Belgium, "*Study of skin changes in cystinosis patients under cysteamine therapy*", Mentor: Elena Levtchenko, MD, PhD. Grant awarded 6 December 2011 for a 12 month period. Total Award: \$99,400.00. This grant is a

joint funding collaboration between Cystinosis Research Network and Cystinosis Foundation Ireland.

Maya Doyle, LCSW, Children's Hospital at Montefiore, Division of Pediatric Nephrology, New York University, "*Cystinosis in Emerging Adulthood*". A one-time grant awarded on 24 July 2011 for a total of \$17,200.

Jess G. Thoene, M.D., Director, Biochemical Genetics Laboratory, Active Professor Emeritus of Pediatrics, University of Michigan, "*Continuation of Feasibility of Cystinosis Replacement Therapy in Cystinosis*", Grant awarded 24 July 2011 for an 18 month period for \$202,500.00.

Rosaleen Anderson, PhD, Sunderland Pharmacy School, University of Sunderland, "*Proteomic investigation of cystinotic cells and the effects of cysteamine treatment*". Grant awarded: 23 November 2010 for a term of 2 years in the amount of GBP 80,926.00 or \$130,366.00 based on the exchange rate for 15 November 2010.

Catherine Tuleu, Ph.D., Ken Nischal, Olufemi Rabi, Rainish Sekhri, Wm Van't Hoff, Univ. of London, School of Pharmacy; "*Development of cysteamine in situ gelling system for the topical treatment of corneal crystals in cystinosis*", Grant awarded January 2007 for 3 years. Total award: 103,000.00 pounds or approximately \$203,500.00.

Leticia Belmont, M.D., Unidad de Genetica de la Nutrician, Instituto Nacional de Pediatría, Mexico. "*Determination of Intraleucocitary Cystine by High Performance Liquid Chromatography (HPLC) in Patients with Cystinosis*". Grant Awarded: 5 November 2008 Total award: \$31,972.00; A presentation, citing CRN funding, was given at the IPNA meeting in NYC, 31 August 2010.

Henk J. Blom, M.D., M.M.C. Wamelink, and E. Levtchenko, M.D., VU University Medical Center, Amsterdam, The Netherlands: "*Newborn Screening of Cystinosis*". Grant Awarded: 5 November 2008; Total Award: 45,000 euros (approximately \$67,500.00).

Francisco Emma, M.D. and Anna Taranta, Ph.D., Bambino Gesù Children's Hospital and Research Institute, Rome, Italy: "*Functional Characterization of Cystinosis-LKG*". Grant Awarded: 5 November 2008; Total Award: \$114,480.00.

Ewa Elenberg, M.D., Texas Children's Hospital, Houston, TX "*Quality of Life in Cystinosis Patients*", Grant awarded 9-25-09 for 1 year. Total award: \$21,000.00

Continued on page 13

CRN Research Update (cont.)

I would like to say a special thank you to everyone associated with CRN, including our most treasured Advisory Boards, and to all of you in the cystinosis community for your support and dedication during the past five years while I have been serving as Vice President Research for CRN.

I first became aware of the Cystinosis Research Network in November of 2004 when my grandson was diagnosed with cystinosis at 17 months of age. CRN was a godsend for me and Mitchell's parents, Eric and Lorna Smith, as, like all of you, we were totally overwhelmed by this never-heard-of-before disease of cystinosis. You smoothed the road for us, and we will be forever grateful.

Colleen Hammond will be returning to CRN's Executive Committee in July, 2012 when she takes over the duties, privileges, and challenges of being Vice President of Research. Colleen has never ceased to be a vital part of this organization since she and her husband, Jack, founded it back in 1996. The cystinosis community is so very fortunate to have her continue to serve us in this capacity.

Respiratory Muscle Weakness a Growing Concern in Adults with Cystinosis

By Richard H. Simon, M.D., CRN Medical Advisory Committee

Muscle weakness is one of the problems that people with cystinosis may experience. Occasionally, the muscles used for breathing are weak and cause symptoms. The most common sign of respiratory muscle weakness is becoming short of breath with physical activity. If someone with cystinosis notices this symptom, he/she should report this to his/her doctor. However, there are many problems unrelated to cystinosis that can cause shortness of breath with activity, so an evaluation needs to be performed. One of the ways to detect muscle weakness is pulmonary function testing that measures the amount of air someone can hold in their lungs and how hard they can suck in and blow out. Additional tests may be recommended including standard x-rays or CT scans of the chest to look for other lung problems. Occasionally tests are done such as an electrocardiogram or an echocardiogram to make sure that heart function is okay and not contributing to the shortness of breath.

If it appears that muscle weakness is causing shortness of breath, supportive treatments are available. When the problem is severe (as measured by symptoms, pulmonary function tests, and blood levels of oxygen and carbon dioxide), it may be recommended that the person use a device to assist their breathing at night and during the day as needed. The device is called BiPAP and consists of a face mask that usually goes over the nose and is attached to a machine that blows air under low pressure to help inflate the lungs. This machine works to insure adequate breathing at night and to rest the muscles used for breathing. An exercise program may also be beneficial. Many medical centers have pulmonary rehabilitation programs that provide guidance in types and levels of exercise that are useful for people with pulmonary problems. To help decide if these treatments are needed, the cystinosis doctor can consult with a pulmonary expert who has experience in evaluating and treating patients with respiratory muscle weakness.

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RP103 Update—Pursuing FDA Review

In March, 2012, Raptor Pharmaceuticals submitted a New Drug Application (“NDA”) for RP103 to the U.S. Food and Drug Administration (“FDA”) and Marketing Authorization Application (“MAA”) to the European Medicines Agency (“EMA”). These applications include efficacy and safety data from nearly three years of clinical trials of RP103 in cystinosis patients in the US and Europe. This was only made possible through the time and effort of all the study participants, families and study coordinators. Raptor greatly appreciates the contributions of all involved in the clinical trials, including many CRN families.

The EMA has accepted Raptor’s MAA and is now reviewing it. Under defined EMA timelines, Raptor expects to receive a decision from EMA in early 2013. As of the publication date of this article, the FDA is performing a top-level review to determine whether they will accept the application for full review. Assuming the FDA accepts Raptor’s NDA for review, an approval decision would be expected sometime between fall of 2012 and early 2013. (Raptor has requested a Priority Review, which may be granted for applications that address an urgent unmet medical need. Under Priority Review, the FDA’s review timeline is 6 months from the date of submission of the NDA. Otherwise, the review timeline is 10 months.)

Continuing Raptor’s Commitment

Since the conclusion of the active Phase 3 study, Raptor has continued to collect data in an ongoing safety extension study, in which patients who completed the Phase 3 clinical trial could choose to continue on RP103 treatment. The goal of the extension study is to monitor safety and white blood cell cystine levels over long-term, steady-state treatment. In order to provide the FDA with data relevant to the younger pediatric population, Raptor also conducted a clinical study showing RP103 can be dosed by sprinkling the capsule contents onto applesauce. The results of this clinical trial enabled Raptor to expand enrollment in the safety extension study to include patients who are too young to swallow whole capsules. Raptor also expanded enrollment in the extension study to include patients who have undergone kidney transplants.

Inclusion and exclusion criteria for participation in Raptor’s clinical studies of RP103 were determined by Raptor and its clinical investigators with input from FDA and EMA, to manage the number of clinical variables and focus on the studies’ primary endpoints in an effort to make RP103 available as soon as possible. It was the responsibility of the clinical investigators at the participating clinical sites, to identify potential patients and determine which patients were eligible for enrollment.

We’re entering an exciting time as we begin the first steps in the FDA review process. Raptor anticipates greater collaboration with the cystinosis community and we look forward to continuing to work with the CRN in our effort to find additional ways to provide even more support to patients and caregivers in the future.

Ophthalmic Cysteamine - Pathway to the Patient



By Karen Kuphal, PhD

Despite the benefits of reducing cystine levels systemically, oral cysteamine (approved by FDA in 1994) is not effective in reducing corneal crystals or the accompanying secondary symptoms of photophobia, blepharospasm, and eye pain (Cantani, 1983; Gahl, 2000; Gahl, 1995). Acknowledging the need for an additional route of delivery, the aggressive development of ophthalmic cysteamine has ensued since its inception in the mid 80's by Dr. Kaiser-Kupfer and her colleagues. The initial experiment conducted in 1985 by her team involved examining the effect of cysteamine in reducing cystine concentration in cultured corneal stroma cells from a patient 13 years of age with debilitating cystinosis. In 1986, she administered the first dose of ophthalmic cysteamine to two cystinosis patients (<2 years of age) over the course of several months (Kaiser-Kupfer, 1986). As a result of the observations in these two patients, a formal clinical trial began that same year, entitled "Protocol 86-EI-0062: A Double Masked Controlled Randomized Clinical Trial of Topical Cysteamine in the Treatment of Corneal Cystine Crystal Accumulation in Cystinosis." This study assessed the tolerability and efficacy of cysteamine ophthalmic solution administered to the cornea of patients who were presently receiving orally administered cysteamine. Over the next several years, additional studies began testing modified formulations at an attempt to improve different attributes of ophthalmic cysteamine (e.g. efficacy, safety, stability, and ease of storage). In all, five formulations aimed at reducing corneal cysteine crystals were investigated between 1986 and 2001 (Kaiser-Kupfer, 1990; Tsilou, 2003; CAPTOC, 2010). Following several amendments and evaluations of the five different formulations, the same clinical trial exists today as "Protocol 86-EI-0062: Trial of Topical Cysteamine in the Treatment of Corneal Crystal Accumulation in Cystinosis." Unlike the design of the original trial, currently enrolled patients receive the exact same formulation that is under review by FDA which was derived from over 19 years of research.

Overall, progress of ophthalmic cysteamine development has come a long way since the work conducted by Dr. Kaiser-Kupfer and several key researchers such as Dr. Gahl, Dr. Schneider, Dr. Thoene, and Dr. Tsilou to name a few. In 2010, Sigma-Tau Pharmaceuticals, Inc. (STPI) submitted to the FDA a New Drug Application (NDA) which included comprehensive clinical-, nonclinical, and manufacturing information relative to the development of ophthalmic cysteamine. Upon review, FDA only had questions about manufacturing facilities of the active pharmaceutical ingredient (API) and finished product. In April, STPI addressed FDA's questions and FDA communicated a new action date of early October 2012 to provide a response. As we await FDA's response, patients can be assured that Dr. Bishop and the NIH remain committed to preserving access to ophthalmic cysteamine through the active ongoing trial (Protocol 86-EI-0062).

Karen is a Sr. Project Manager at Sigma-Tau Pharmaceuticals, Inc. She may be reached at Karen.Kuphal@sigmatau.com

References

Cantani A, Giardini O, Ciarella Cantani A. Nephropathic cystinosis: ineffectiveness of cysteamine therapy for ocular changes. *Am J Ophthalmol*, 1983;95(5):713-714.

Combined Analysis of Patients Treated with Ophthalmic Cysteamine (CAPTOC); Clinical Study Report STP869294 on file at Sigma-Tau Pharmaceuticals, Inc.

Gahl WA, Kuehl EM, Iwata F, Lindblad A, Kaiser-Kupfer MI. Corneal crystals in nephropathic cystinosis: natural history and treatment with cysteamine eyedrops. *Mol Genet Metab*, 2000;71(1-2):100-120.

Gahl WA, Schneider JA, Aula P. Lysosomal transport disorders - Cystinosis and sialic acid storage diseases. *The Metabolic and Molecular Bases of Inherited Disease*. New York, NY; McGraw-Hill;1995.

Kaiser-Kupfer MI, Fujikawa L, Kuwabara T, Jain S, Gahl WA. Removal of corneal crystals by topical cysteamine in nephropathic cystinosis. *N Engl J Med*, 1987;316(13):775-779.

Kaiser-Kupfer MI, Gazzo MA, Datiles MB, Caruso RC, Kuehl EM, et al. A randomized placebo-controlled trial of cysteamine eye drops in nephropathic cystinosis. *Arch Ophthalmol*, 1990;108(5):689-693.

Tsilou ET, Thompson D, Lindblad AS, Reed GF, Rubin B, et al. A multicentre randomized, double masked, clinical trial of a new formulation of topical cysteamine for the treatment of corneal cystine crystals in cystinosis. *Br J Ophthalmol*, 2003;87(1):28-31.

CRN Education and Awareness Update

Upcoming Family and Medical Events

April 28-May 1, 2012

2012 PAS/ASPN Annual Meeting
Boston, MA

May 15, 2012

NORD Partners in Progress Gala and Corporate Council Meeting
Washington DC

August 15-17, 2012

Cystinosis Mexico Symposium
Mexico City, Mexico

Oct. 30-Nov. 4, 2012

2012 ASN Renal Week
San Diego, CA

May 3-7, 2013

2013 PAS/ASPN Annual Meeting
Washington DC

July 18-20, 2013

CRN Family Conference
Washington DC

By Paula Shal-Vice President, Education & Awareness

I struggled to write the E&A update for this newsletter. After eight years, my term on the CRN board ends in July, and this is the last newsletter I will edit. So emotions ran deep as I decided just exactly how to write my "swan song."

I decided I am not going to write about the impact CRN has made at the ASN and ASPN meetings over the last few months as we educated doctors from all over the world about cystinosis and made a direct impact on doctors with limited access to cystagon in other countries. Nope, why would I mention that? And I am not going to write about how CRN Board member Jenni Sexstone as well as Roison Keon exhibited at meetings for the first time. I also won't mention how CRN had a presence in Washington on Rare Disease Day February 29.

I won't even discuss the tears I shed while reading the essays and recommendations of the three CRN Scholarship applicants and how gut-wrenching difficult it was to choose one winner ...so we chose all three. Nope, I am not going there.

And I won't write about the incredible young adults with cystinosis who are graduating from high school and college and how the number of college graduates with cystinosis is increasing.

Okay, maybe I will write about that a little. As more and more individuals with cystinosis emerge into adulthood, adult cystinosis issues have become more prevalent, and there is a desperate need for research for these issues. As an adult with cystinosis, I have experienced many of the complications adults face. I was not diagnosed with cystinosis until I was 17 years old. I was told I may have a "mild form" of cystinosis, and my only symptoms as a child were photophobia and stunted growth. Of course, I was not being treated at all with cysteamine since no one knew I even had cystinosis. The lack of treatment took its toll on my muscles and at age 17, it was clear they were starting to atrophy. Today, at age 41, I have significant muscle wasting in my hands, feet, respiratory system, facial muscles, vocal folds, and just overall.

For me, muscle wasting is the single most challenging part of living with cystinosis—more challenging than the nine doses of medicine I take around the clock each day. More than the occasional "digestive upset" that comes with it. More challenging than the four finger pricks and four insulin shots I need each day due to steroid-induced diabetes. More challenging than photophobia and eye drops. More challenging than a kidney transplant (piece of cake!) And it rivals my experience with dialysis. Muscle wasting is a challenge 24/7. Physically, it is a challenge every time I swallow. Every time I speak. Every time I use my hands. Mentally, it is a constant reminder I have a disease that is expected to shorten my lifespan.

To the parents of kids and teens with cystinosis, if you need an example of what can happen if your kids don't take their cystagon, feel free to use me. If you need some shock value, tell them I rarely text or use a cell phone because I don't have the fine motor skills to efficiently operate one. I don't mind not having this technology. Really. But they might. You can also have them wear mittens all day and try to do normal tasks. The hand functionality with a pair of mittens closely represents the functionality of hands that are severely atrophied from cystinosis. By some miracle, I have retained the muscles which help me type. Without this ability, having a career (and being on the CRN Board) would be much more challenging.

It was thought that cysteamine therapy from a young age might divert muscle wasting all together. But as some adults who have been compliant a majority of their lives emerge into their upper twenties, they are starting to show some signs of muscle wasting. The good news is that there is a great "buzz" in the cystinosis community about muscle wasting in adults. Our medical community sees what healthy, productive members of society adults with cystinosis are and are determined to provide better treatments for all aspects of this complex disease. I see hope in our future. But we must press on.

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Paula and Jim Shal—Pickle, Buzz, and Keeper

CRN Scholarship applications now available!

See page 30-31 for details.

CRN Education and Awareness Update (cont.)

There are currently five adults with cystinosis on the CRN Board, and I cannot wait to witness how they and the rest of the board continue to make progress regarding issues that impact adults. One of our board members with cystinosis, Jessica Jondle, will assume the responsibilities of VP-Education & Awareness. Jessica brings a great deal of knowledge about cystinosis as well as creativity, writing, editing, and technical skills to the board. Jessica has already written a book and is an inspiration to our community. I am so excited to see how the newsletter evolves with her as editor and to see how the Education & Awareness Committee thrives under her leadership!

I have thoroughly enjoyed my eight years on the board. I have made some great lifelong friends, and I hope I have contributed in some way and made a difference in the lives of families and individuals living with cystinosis. Being on the CRN board has taught me to learn more, do more, and be more. I have volunteered for one-time events and volunteer tutored on a weekly basis, but I have never pushed myself as much as I have as a CRN board member. I believe in CRN's mission and vision, which was the fire I needed to keep going.

My time on the board wasn't always easy. I worked 45+ hours a week in a demanding job. I was on dialysis for five months and ultimately had a transplant. I had so many UTIs, fevers, bowel obstructions, and aspiration pneumonias that landed me in the emergency room, I lost count. My health complications were a good reminder of why I originally signed up to be on the board—so that future generations of people with cystinosis do not have to face the same challenges I did. Rather than let go of my responsibilities on the board, there were so many times I wished I didn't have to work at my paying job and could just focus on CRN. The board has so many great ideas but not always the resources to implement them. I wished for the time to bring these ideas to fruition. *But then reality would sink in...*

My husband and I both work in unsteady corporate America. We know that if one of us loses our job, we have to rely on the other's health insurance. It is important for us to both keep working. Everything we have worked so hard for could be gone in a flash with my prescription drugs alone. I know there are many out there who feel my pain. So I plug along putting 110% into my job with the hopes of reducing my chances of losing it...and I think about winning the lottery.

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I was a healthy baby who could "flip the bird"



Not a typical toddler with cystinosis—I liked to eat, and it showed.



Only signs of cystinosis as a child were short stature and mild photophobia



Giving my friend rabbit ears—haven't been able to do this for awhile



Was in renal failure as a freshman in high school but didn't know why



A senior in college and two transplants under my belt



Day of discharge after 3rd transplant—age 31 and unable to straighten hands



With my friend and kidney donor, Sue, after 4th transplant taping a news spot

Report from the National Organization for Rare Disorders (NORD)



By Marybeth Krummenacker—NORD Board of Directors, CRN Board of Directors

The National Organization for Rare Disorders has been working extraordinarily hard at keeping pace with so many facets of the rare disease community. As I said in my last update, we live in a face-paced world and people are looking for instant answers and quick fixes to all issues concerning rare diseases. From diagnosis and the latest research to innovative discoveries and new medications, it is important to be aware of all of the issues that could affect those of us living with a rare condition such as cystinosis. Again, I am honored at this incredible opportunity, and I have tried to represent cystinosis the best that I can on NORD's Board of Directors and to be the best advocate for the cystinosis community as a whole. After living with this disease for more than 23 years, there is still much to be learned!

NORD's responsibility to the rare disease community is enormous. The organization has held two additional regional meetings since the meeting in Chicago in September 2011, one in New York City in December and one in San Francisco in March. NORD has a renewed commitment to continue to hold these regional meetings over the course of the next year; dates and locations are being discussed now and will be announced over the next few months. These meetings have been hugely successful and thought provoking. The staff has gotten opportunities to meet with the advocacy groups throughout the country and get a true sense of what it is they are looking for from NORD. The Membership Committee (which I have been asked to Chair) has received double the amount of membership applications than were received by this time last year! This is as a direct result of the regional meetings.

NORD's networking ability and personal contacts have helped to raise NORD's voice. The relationships that continue to be developed with the NIH, FDA, SSA, and Capitol Hill are critical to moving forward with an agenda for rare diseases. The recent Rare Disease Day events held at the NIH and the FDA were outstanding. CRN was fortunate to have representation there to network with the leaders of both government agencies. From all accounts, Rare Disease Day 2012 was a huge success and one that will be built on for next year. As this newsletter is going to press, NORD is preparing for its annual Gala in May. This year's event will be held at historic Union Station and will once again have representatives from CRN in attendance.

Finally, I would like to leave you with something I recently read, written by President Theodore Roosevelt:

*"It is not the critic who counts; not the man who points out how the strong man stumbles, or where the doer of deeds could have done them better. The credit belongs to the man **who is actually in the arena**, whose face is marred by dust and sweat and blood; who strives valiantly; who errs, who comes short again and again, because there is no effort without error and shortcoming; but who does actually strive to do the deeds; who knows great enthusiasms, the great devotions; who spends himself in a worthy cause; who at the best knows in the end the triumph of high achievement, and who at the worst, if he fails, at least fails while daring greatly, so that his place shall never be with those cold and timid souls who neither know victory or defeat."*

With respect to the tremendous work done by the National Organization for Rare Disorders, the CRN continues to be a presence "in the arena" on behalf of the cystinosis community.



CRN is a proud sponsor of this year's symposium!

CYSTINOSIS MEXICO
ANNOUNCES:



6th Cystinosis Symposium
Mexico City

August 16-17, 2012

More information coming soon

www.cystinosismexico.com

Cystinosis Foundation Ireland Update

There is exciting news on the research front, with Cystinosis Foundation Ireland and the Cystinosis Research Network embarking on a funding collaboration. The project we are jointly funding is entitled "**Study of skin changes in cystinosis patients under cysteamine therapy**" with Martine Besouw, M.D. as the Principal Investigator. This is a major step forward for our organizations and for the future of research internationally, and we are very pleased to be a part of this.

In addition to this collaboration, Cystinosis Foundation Ireland is currently working on the MRCG/HRB Joint Funding scheme and reviewing quite a few research projects to be considered for funding. The MRCG is the Medical Research Charities Group and is an umbrella organization for medical charities in Ireland. The HRB, Health Research Board, is the Irish Government's scientific funding body. Under this scheme, the MRCG/HRB and the charity split the cost of the research project 50/50. This year each charity will only need to contribute 25% to any successful projects.



At present, this year's projects are being peer reviewed before our Executive Board will meet and decide which, if any, we will fund. It is always very pleasing for us when new research projects, and especially new researchers, come in contact with us as this offers fresh hope, excitement, insight, and ideas into the world of cystinosis research.

As always, we are constantly fundraising and thinking of new ways to tempt people to get involved. We have a few events in the pipeline this year and we will let you know in the Fall newsletter how they went!

Follow us!

Facebook: <http://facebook.com/cystinosis.ireland>

Twitter: @CystinosisIrl

Web: <http://www.cystinosis.ie>

Cystinosis Story—The Taylor Family



Peytan, Jamie, and Channing Taylor

Hello Everyone! We are the Taylors and we are the proud parents of a 2-year-old little girl named Peytan Preslie Taylor. Our names are Channing, 24, Jamie, 27, and Peytan, 2. Jamie and I have been married for 1 year in May, and have been together for about 4 years now. We live in Chesapeake, Virginia, and love it! I am a stay-at-home mom and my husband is an EMT for the City of Virginia Beach.

Peytan was diagnosed with Cystinosis in February 2010. We had no clue what Cystinosis was, so we were completely floored when her doctor came into the room at a regular check-up and informed us of this rare disease. Peytan was about 1 year old when she was diagnosed. I thank God everyday that the doctors caught this disease when they did!

The only symptom that we noticed Peytan had before we knew what was wrong was that she wasn't growing or gaining weight. In fact, she had lost weight. I wasn't aware of the difference in her facial structure because I saw her every day. My dad and Jamie's mom were the ones who mentioned that we should have her checked out, because they felt something was not right. She had been growing and gaining weight naturally and normally up until she was 1 year old. After tons of blood work, and seeing an endocrinologist, her pediatrician, as well as her nephrologist, the doctors finally figured out what had overcome our little girl's life, as well as ours. Since Peytan - like most children with cystinosis - was not a big fan of food, she lost her means to chew and became funny about different textures. That caused her to lose weight even more. Peytan's eating habits are still very minimal. She eats macaroni and cheese and chicken nuggets. That is what she craves and what she likes to eat! Her appetite is strong for two weeks and then it dies off for about two weeks. We have her in occupational therapy to help her get her appetite back, learn about different textures, and be able to chew anything. Peytan takes her meds every 6 hours, gets her growth hormone shot once a day, and gets eye drops every hour while she is awake. The medicines that Peytan is taking are Cystagon, Calcitriol, Potassium Citric Acid, Sodium Citric Acid, Levocarnitine, Levothyroxine, Zantac, Eryped, Zofran, Prilosec, iron, and Ranitidine, along with the Nutropin AQ and cysteamine drops for her eyes.



Peytan Taylor

Peytan is so playful that she is ready to go by 7 in the morning! Our daily routine consists of playing, learning, crafting, medicines, eye drops, eating, shot, and then bed. She definitely keeps us entertained and on our toes with her wittiness and huge personality. Peytan is the bravest person that we know. Someone who has just met Peytan would never imagine she is fighting this life-long battle. She shows no struggle with cystinosis and is full of love and life. Peytan is the happiest, most fun-loving child that we have ever met, and we are so blessed that we get to call her our daughter! We are coping and adjusting to cystinosis pretty well. My husband is perhaps coping better than I am, but I know we can't let this disease run our lives. We can't shelter Peytan and make her scared of this disease because we

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Cystinosis Story—The Taylor Family (cont.)



Peytan Taylor

will find a cure one day, and she will beat it! We have to be hopeful and optimistic, or we would never be able to give the full support and love we do to Peyton. Our parents adjusted to this astonishing news fairly well and are always there to help us when we need it. My parents help us out tremendously with her medicines, holding her so I can give her shot to her, as well as giving her eye drops while I am doing other things for Peyton.

The biggest challenge and adjustment Jamie and I had to face was the realization that our little girl has this disease and will have it for the rest of her life. It was also hard getting used to having to do the meds, shot, therapy, pump, doctor appointments every two weeks, and eye drops. It is extremely frustrating that I can't take Peyton's place and let our little girl have the life she deserves. This disease has brought us closer as a family because we need to lean on each other.

The really big adjustment that Peyton had to get used to was the medicine and the shots. She does amazing at everything that she has to go through; she absolutely loves her doctor, Dr. Raafat, and all of the ladies in her office! Peyton acts like a normal toddler and is full of life. She is the sweetest, most loving, and kind-hearted little girl that you will ever meet. She has a

bigger-than-life personality and has a smile that will light up a room and make you laugh when you cry. She truly is the light of our lives and has changed us as people and as parents. Because of her, I am a better person.

CRN has been a huge support system for us. The organization has answered questions, comforted us in times of despair, linked us to the NIH to see Dr. Gahl, and welcomed us into the cystinosis family. We are absolutely ecstatic to be a part of such a close group of people! Our hopes and dreams for Peyton are that she will get to live as normal of a life as she can, that she can tackle and achieve all of her hopes and dreams, and that she will be able to live her life to the fullest.

Cystinosis Story—Jordan Darling



Jordan Darling

My name is Jordan Darling, and I am from Texas. I was born On May 10, 2006 I was 8lbs 12 oz. I had to be care flighted to Medical City due to water on my lungs and trouble breathing. I stayed here for a little over a week and was released to go home when I could breathe on my own. I did real good up until I was about 9-10 months old when my mom noticed that I wasn't gaining weight and wasn't even trying to walk. My mommy took me in to see my pediatrician, and he wanted me to go see a genetic specialist at Children's in Dallas! The specialist had me admitted into the hospital for further testing. Little did we know it was going to be a very long stay! After many tests and research we were told that I had Cystinosis! My life would be changed forever!

I am in kindergarten, and I love school! I also love wrestling, playing outside, and the Dallas Mavericks.

Cystinosis Story—Landon Hartz



Landon Hartz

Landon was born on April 8, 2011 weighing 7lbs, 4oz and was thought to be perfectly healthy. He was never a big eater, but was growing well for the first 6-7 months of his life. Then his pediatrician noticed that the growing slowed down and then stopped around 9 months. She coached us on how much formula he should be consuming. He seemed to enjoy solids for a while but then around 10 months, even seemed less interested in his food. We were referred to a gastroenterologist who could find nothing medically wrong so ordered several tests to check for a variety of GI related issues as well as Cystic Fibrosis. We were referred to a nutritionist and feeding team and although they made wonderful recommendations, nothing worked. Then, Landon began vomiting. We were sure that he had acid reflux and if he had silent reflux, not wanting to eat would make sense. The gastroenterologist ordered a scope right before Landon turned 14 months. Again, she found nothing. She decided to admit him to Children's Hospital of Pittsburgh for an intense feeding therapy on June 2, 2011. Her hope was that by having a team to work on feeding issues with him on an inpatient basis, that we might figure out why he's not eating and work on a solution.

When Landon had the scope done, the gastroenterologist decided to collect a urine sample. When she called to give admission instructions, she indicated that sugar was found in his urine but that it was probably a fluke. She ordered another urinalysis upon admission to the hospital. As a result of this urinalysis, a renal doctor was sent in to talk with us. Landon was diagnosed with Fanconi Syndrome. He explained that sometimes Fanconi Syndrome stands on its own, but often times it is a result of a metabolic disorder. After reviewing all of the possibilities, a rare disease called Cystinosis made the most sense. One week later, the blood test that was sent to a laboratory in California indicated that our baby did, in fact, have this rare disease called Cystinosis. Landon was hospitalized for 25 days before we were sent home, our lives forever changed.

Challenges encountered to date

Landon and our family have faced multiple challenges since he was diagnosed. Learning and accepting our new "normal" is something that we continue to strive towards. Landon is so resilient and amazes us every day.

Landon is fed, at this point, mainly from a feeding tube. He gets bolus feeds throughout the day and a continuous feed for 10 hours at night. For the first six months, we struggled off and on with vomiting. We experimented with how much his belly can handle at once, whether we should slow the feed down or can we speed it up a bit, should we do 4 small bolus feeds or 3 and add an extra ounce and so forth. In the past month, it seems like the vomiting has become much less frequent.

Another challenge we have faced is getting him to take in food orally. We have been consistent about offering him food eat each meal, right before a feed, and we have initiated services with an early intervention group. An occupational therapist comes to our house once a week to work on feeding issues. For a few months, Landon showed very little interest in eating. In time, he has developed interest and asks to eat all throughout the day. Another obstacle that we encountered, however, is that he holds food in the side of his mouth and doesn't swallow it. Again, with time and some exercises, he is beginning to take in more and actually swallows. The feeding tube is not going away any time soon, but we are certainly making progress.

Adjusting his medication has also been a bit of a challenge from time to time. It is important to keep his electrolytes within a normal range so he has to have blood taken regularly to see how everything looks. We have been fortunate in that Landon has tolerated his medication pretty well and his levels look pretty good for the most part, but there certainly has been some experimentation involved with that.

Continued on page 23

Cystinosis Story—Landon Hartz (cont.)

His gross motor skills were also a concern for us. Landon was on target for most skills except for walking. We also initiated early intervention services with a physical therapist to work on this. We are proud to say that our little guy is walking now and has made incredibly progress in the past few months and we continue to work with the physical therapist to support him.

How has the diagnosis affected us

Cystinosis absolutely rocked our world, but we were thankful for answers. At the beginning, it seemed as if we went through all of the stages of grief over and over again and sometimes all in the same day! We still go through some of those stages at times, but the bad days seem to be less and less. It has made us feel more vulnerable. If this can happen to our child and our family, anything can happen. On the other hand, we have found strength in ourselves and in each other. We are learning how not to let Cystinosis consume our lives. We are involved in fundraising and we have connected with hundreds of people who are also affected by this disease, but Landon is much more than Cystinosis. He is a normal little boy that has some special needs.

Typical day in the life of Landon

Landon usually wakes around 8:00am. I wake one hour before to give him a medicine called Levothyroxine which is used to keep his thyroid levels in a normal range. He was recently diagnosed with Hypothyroidism which we learned is common in people diagnosed with Cystinosis. All of Landon's medications are given through his Mic Key button. At 8:00am, I start his first bolus feed of the day which usually takes about one hour from start to finish. Around this time he also takes his first day time dose of Cystagon along with three other medications. Depending on the day, we may leave after this feed and he goes to "school" which is a Mommy and Me pre-school program that the two of us go to together once a week or we run errands, visit friends or grandparents and sometimes just stay in the house and play. Around 11:00am, we have lunch. Some days he eats a little bit and other days he just plays with the food and won't actually eat anything. At 12:00pm, he goes down for a nap and starts his second bolus feed of the day. He also gets three more medications at this time. He typically naps until around 2:00pm and when he wakes, it's time for Cystagon and one other medication. After his nap we may run errands, visit friends or family and two days a week he has either occupational therapy or physical therapy. At 3:30pm, Landon is given something to eat and at 4:00pm, we start his third bolus feed of the day and he takes four medications. The rest of the evening includes dinner for Mommy and Daddy while Landon sits in his high chair and participates in dinner with us, play time, bath time, stories and bed at 7:00pm. His feed starts at 7:00pm and he is given three medications and then at 8:00pm, he is given Cystagon and one other medication. At 2:00am, I wake to refill his formula and to give him Cystagon along with one other medication. He stays asleep during this time.

During the feeds in which he is awake, we spend time watching the television shows that he likes, doing puzzles, playing with toys and reading books. His feeds and medication schedule never stop us from going anywhere or doing anything. The feeds and medication schedule was a bit overwhelming at first, but now, it's just what we do. We do not think much about it anymore.



Jimmy, Lauren, and Landon Hartz



Landon Hartz

CRN Development Update

By Jen Wyman-Vice President, Development



Jen and Kacy Wyman

"We must not, in trying to think about how we can make a big difference, ignore the small daily differences we can make which, over time, add up to the big differences that we often don't foresee."

Morian Wright Edelman

Our daughter Kacy, age 9 with cystinosis, wakes up EVERY SINGLE DAY asking "What do we have today?" Now I know when she is asking that question she isn't referring to anything "cystinosis". She is referring to her social life and swimming and dance. She is referring to school and play dates and weekend activities. Thankfully she is looking forward to each and every day and what it has to offer her because of what we have today in terms of medications and treatments and surgeries. We have much to be grateful for on that front as well! Within just a couple of decades we have gone from no treatment to improved treatments. We have multiple doctors doing multiple research projects. We have gone from the diagnosis of cystinosis as a death sentence to witnessing moms with cystinosis delivering healthy children, to becoming authors, to living their lives decade after decade after decade.

Sixteen years ago one family had a child with cystinosis. Sixteen years ago one family had one idea to start the Cystinosis Research Network; to unite families and physicians in the cystinosis community with the common goal of providing the best quality of care to everyone living with cystinosis. And sixteen years later the Cystinosis Research Network is raising hundreds of thousands of dollars to fund research projects for dedicated doctors. We are raising hundreds of thousands of dollars to hold conferences all over the country for a few hundred people living with cystinosis. Without facebook and email and the internet one family made a difference that we are all reaping the benefits from today. Thank you Hammond Family for this gift.

SO...what do we have today? Let me tell you...we have a dynamic network of individuals from all walks of life and from all over the world who are affected by cystinosis, and linked together through social networking. We have medications that are improving the quality of life and increasing our loved ones chances of living long healthy lives. We have role models to live by and information at our fingertips. We have a cystinosis family that we can lean on and learn from in times of need.

What do we have today? We have families holding fundraisers year after year...bringing in much needed money to fund much needed research to continue the path we are on. This year was no different...we had many families who held fundraising events in every capacity. No matter the size, each touched the lives of many, and each contribution was as important as the next. Since 2012 began the Krummenaker/Maccarone families held their dinner dance, the Woodward Family is planning their 6th annual golf outing and the Larimore Family embarked on their first golf outing. The Greeley and Schleuder Families wrote letter campaigns, Jessica Jondle authored her first memoir, donating the proceeds to CRN and my family held our 6th annual Fun Run. Individually each of these fundraisers brings in big dollars. Collectively they touch multiple people from all over the world...spreading knowledge and information to hundreds of thousands of people who otherwise

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CRN Development Update (cont.)

may never hear the word cystinosis, let alone have an understanding of what it is or ever meet a person who lives with this disease.

Perhaps if you ask yourself "what do I have today" you will find yourself embarking on new adventures or just appreciating the little things you do each and every day. Perhaps you will ask yourself what can I do for my child, or my spouse, or my parent that is living with cystinosis today? It doesn't have to be big to be meaningful and it doesn't have to be complicated to be worthwhile. It can be small and simple and special. "What you do today can improve all your tomorrows." (Ralph Marston)

"To get up each morning with the resolve to be happy... is to set our own conditions to the events of each day. To do this is to condition circumstances instead of being conditioned by them."

Ralph Waldo Emerson

2012 Fundraising Calendar

Date	Event	Location	In Honor Of
February 13	Steve Schleuder Birthday Letter	Novi, MI	Steve Schleuder
March 13	Milleridge Dinner Dance	Milleridge Cottage Jericho, NY	Laura Kruppenacker The Maccarone Family
March 17	Jack Greeley Birthday Letter	Lincolnshire, IL	Jack Greeley
March 23	Swings for Sarah Golf Event	Blythewood, SC	Sarah Larimore
April	Gabe Stephenson Birthday Letter	Kentucky	Gabe Stephenson
May 6	6th Annual 5K Fun Run	Bloomfield Hills, MI	Kacy Wyman
June 9	6th Annual CH Robinson Golf Tournament	Lehi, UT	Tahnie Woodward
June 15	Sarah Larimore Birthday Letter	Blythewood, SC	Sarah Larimore
September	Kacy Wyman Anniversary Letter Campaign	Bloomfield Hills, MI	Kacy Wyman
October	Sakat's Manor Haunted House	Gettysburg, PA	Laura McGinnis
Ongoing	Jessica Jondle Book Fundraiser	Amazon.com bn.com	Jessica Jondle

10th Annual Jack Greeley Birthday Letter



Great Grandpa Arndt, Christy, Jack, Alex and Dave Greeley celebrate Jack and Great Grandpa's birthdays

For the tenth year, the Greeley family has sent their annual appeal to friends, family and colleagues to raise money for CRN in honor of their son Jack's 12th birthday on St. Patrick's Day. Proceeds raised from this year's letter will push the total donation to CRN to nearly \$200,000. Below is an excerpt from this year's letter. The letter in its entirety can be found on the CRN website at www.cystinosis.org/filemanager/file/Fundraising%202012/JackCRNletter2012%20FINAL.pdf

"Watching Jack eat dinner is kind of an event for us...part celebration and part comic relief. First of all, when your kid does not eat for 18 months, it becomes a simple pleasure to watch him actually do it. To see Jack (who we think is part caveman with his love of meat) dunk his carne du jour in a mound of ketchup is pure nirvana. For years, Jack was skin and bones, but over the past year or more, he has actually put some extra meat on his bones from his love of eating to the point where he is actually a little chubby. Dinner time also gives us laughs because Jack is a SLOW eater. When he was terribly ill, he found his "happy place" in front of the TV – watching Bob the Builder, Power Rangers, you name it. He helped self medicate and soothe himself when the TV was on, enabling us to coax some medicine into his body. That likely lurks in Jack's memory to the point now where we think he has crafted a masterful strategy where if he is so much slower than anyone else eating, Mom, Dad, and sister will get dinner fatigue and leave the table, thus allowing Jack to finish his dinner with the TV on back in his "happy place." The kid is dumb as a fox!



On St. Patrick's Day, Jack's 6th/7th/8th grade basketball team won the house basketball championship...a great birthday present.

Jack's development hopefully reminds anyone who knows him that each day is a blessing. While the world seems to get more chaotic every day, a daily feast awaits everyone who can bring perspective to their life. Compared to ourselves, we all know that someone has more of this or less of that...richer or poorer, smarter or dumber, more or less attractive, happier or sadder. Life serves us all something different and it will not be the same thing tomorrow that it is today. Is your life too bland, too spicy, too salty, or too sweet? Everyone has an opportunity to determine how they want to season what they get served. Yes, if life gives you lemons, you should consider making lemonade. Charles Swindoll reminds us of this when he said: "The longer I live, the more I realize the impact of attitude on life...we have a choice everyday regarding the attitude we will embrace for that day. I am convinced that life is 10% what happens to me and 90% how I react to it."

Jack reminds us all of Swindoll's wisdom. He very rarely complains and he has never lamented having Cystinosis. Alex underscored this recently when she was assigned an essay to write about her hero. She chose Jack and if given the opportunity, Christy and Dave would make the same choice. For us, Jack is the cherry on top of our sundae...the chocolate chips in our chocolate chip cookies. We have been extremely fortunate to have a caring family and so many wonderful friends to share our stories with over the years and we have received incredibly generous support in return. With this on-going fundraising letter writing campaign, we have been able to raise around \$165,000 to help battle Cystinosis. It has been humbling for us and we thank you all."

The Greeley family is thankful for the support of so many generous friends and family and a community that has supported them so enthusiastically over the years.

Steve Schleuder's Birthday Letter

By Terri Schleuder

February 2012 seemed like the perfect time to hold a fundraiser for CRN. February 13th was our son, Steven's, 24th birthday, and February 29th was Rare Disease Day. Bringing awareness to the fact that 7,000 rare diseases affect 30,000,000 people in this country while highlighting Cystinosis through Steve's story seemed like a win/win.

We chose a birthday letter format that emphasized the amazing progress in Cystinosis research over the last 35 years. We described the mission of the Cystinosis Research Network to support families, provide educational awareness to the medical community and others, and to fund promising research projects that offer hope for improved treatments and ultimately a cure for those afflicted with cystinosis. We also discussed the need for continued research efforts in many areas including addressing the needs of some in the emerging adult population of cystinosis patients who have been compliant with cystagon since diagnosis and are beginning to experience devastating late symptoms.

We sent out our letter to family, friends, those on our Christmas card list, select e-mail addresses and to both of our groups of co-workers. (We got permission ahead of time to include co-workers.) Several of those people forwarded it on to others. The response was phenomenal. To date the effort has raised \$9,263.00 with more money continuing to come in. We continue to be overwhelmed, and humbled by the show of support from so many people, some of whom we've never even met.

For me, the decision to fundraise was not an easy one. I felt fear, bordering on terror at the thought of asking people for money even for a cause as dear as this one is to us. One statement Christy Greeley made in a recent newsletter kept replaying in my head, "...If not now then when?... If not me then who?" It was enough to get us started. After writing the letter we mailed it off and asked God to deliver the results. We have been amazed by the response. My unfounded, initial fears about fundraising melted away under the power of the compassion, friendship and support of so many wonderful generous people.

Our family has always believed there is good to be found even in the most challenging of situations. The good we have found because of Cystinosis in our lives continues to shine through the people we've met, the opportunities we've had, and the love and support given to us constantly and unconditionally over the last 24 years. We are truly blessed.

For anyone thinking about holding a fundraiser, don't be afraid to jump in. The water really is fine. And the results will astound you.



Carl, Steve, and Terri Schleuder

Did you know?

There is a comprehensive Cystinosis Article Library on the CRN website at www.cystinosis.org/faq.

There is a list of doctors who treat cystinosis patients on the CRN website at www.cystinosis.org/what-is-cystinosis/resources/doctors.

You can connect with families in your area by registering your contact info at <http://www.cystinosis.org/families-in-your-area>.

Sigler Family Inspires at Milleridge Fundraiser

The Miracles at Milleridge dinner/dance fundraiser took place on March 10, 2012 at Milleridge Cottage in Jericho, NY. The event, which has become a tradition, raised over \$17,000 and was another fun-filled evening featuring dinner, dancing, raffles, and a silent auction.

The highlight of the evening was when Marybeth Krummenacker shared the story of Jessica and Herberth Sigler from Columbia, South America. The Siglers' daughter, Martina, has cystinosis. The family has had an extraordinary journey in their struggle to find proper treatment for Martina. They are currently living at the Ronald McDonald House in New York City, and Martina is being seen by Dr. Rick Kaskel at Montefiore Hospital in the Bronx, New York. The family was able to spend the evening at the fundraiser. They were driven to and from the event in a limousine (thank you John Maccarone) and were overwhelmed by the kindness of people speaking with them. It really was a beautiful story to tell to the families gathered to support CRN. A special thank you to the Maccarone and Hammond families for your help and support and in particular to the Krummenacker and Murphy family and friends for ALWAYS being so supportive. Another successful event and evening!!!



John Maccarone, Herberth Sigler, Jessica Sigler, Martina Sigler, Shea Hammond, Colleen Hammond, Arlene Maccarone, Laura Krummenacker, Marybeth Krummenacker, Jack Hammond at the Miracles at Milleridge dinner dance fundraiser.

T-Shirt Sales Raise Money for CRN



H
Garrett Amelia Glenn Ami Jessica Devin Nevaeh
Aimee Ray Jeffery Ken 7/8/10 Josie Abbi Lucy Evan Euan Nelly
Jacqueline 7/24/98 Luka Devlin Jakob Emma Joyla Graziella Kacy Briana Camyll Lavie Kiera Brooke Kirsty Jason Marcus Lucas Missy Gracie Regina 10/14/73 Tom G
Andrew Maddi Mitchell Joshua Serena Shahrazad Tina Tom K

O
Harley James Anjie Emilie Arrabella Jennifer Bryan HarryBoy Chance 11/8/76 Samantha B Eddie Alex Briar Morgan P Morgan F Erin 8/17/02 Paula Edgar Anthony Landon Luis Armando 5/26/07 Jeff Joel Patrick H Alexia Patrick M Elliot Erik Roxanne 10/30/63 Ryan Shea Sam Channing Tom M Siera

P
Samantha V Heidi Kalob Fabian Emily P Camden C Kate Aliyah Mitchell 10/9/05 Laura Fidelity Sarah E Holly Ida
Austin M Lincoln Kayla Dakota Camden L Michael 1/27/64 Lesley Christina Kelly Alicia
Mason Matthew

E
Chad Emily A Livia Steph Bailey D Sophie Jorjin Zac Christopher Amber 8/2/10 Kennedy Bill J Mack Sarah L Jackson Bailey P Jacob Jessica C 1/3/06 Jordan Kimberly Gabbie Daniel Sky Emily M Kelli 4/12/05 Mikaela Amanda Sherita 10/11/88 Camden S Sarah M Jack G Poppy-Mae Wyatt Steve Whitney Jack K Yago Bree

Micke Newcomb had a vision to bring the cystinosis community together for a common goal. Born from that goal was a t-shirt with the graphic "Hope has many names" on it with the names of children and adults with cystinosis inside the letters in the word HOPE. Those who are deceased were also included.

Sarah Melang was instrumental in creating the graphic for the shirt. Dena Gard, Doug Miller, Catharine Kelly, Bryan Stout, Lynn Thomas,

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Birdies & Cheers Abound at Inaugural “Swings for Sarah” Golf Event & 19th Hole Celebration

The inaugural Swings for Sarah golf outing and 19th hole celebration benefiting the Cystinosis Research Network was held on March 23rd. The event was presented by the Larimore family on behalf of Sarah Larimore and was held at the Golf Club of South Carolina at Crickentree in Blythewood, S.C.

Sixty-four players teed up in the golf event and guests travelled from as faraway as Charleston, S.C. and Charlotte, N.C. to participate. Another round of guests joined in the post-round raffle drawings and all were provided a catered buffet of fine Southern cuisine and desserts.



A good number of eagles and birdies were made during the afternoon as the Carolina weather of warm sunshine and light breezes was perfect for low scores. The handicap-adjusted score of 51 carded by the team sponsored by Palmetto Payroll Solutions of Columbia, S.C. was the winning score.

The Larimores would like to extend gratitude to all of the players, Friends of Sarah sponsors, and the corporate sponsors from Charley’s Grilled Subs, Gateway Supply, Thunder Tower Harley Davidson, & Carolina Comfort, plus Bauknight Pietras & Stormer CPAs for their contributions and giving this inaugural event the momentum to make the Swings for Sarah 2013 event a must for everyone’s calendar.



Sarah Larimore

Swings for Sarah raised over \$6,300 for CRN!

Team Kacy! 6th Annual Cystinosis Fun Run

By Jen Wyman

Sunny skies and smiling faces surrounded us as over 300 gathered, once again, for the 6th Annual Cystinosis Fun Run honoring our daughter, Kacy! It is my favorite day of the year...when those closest to us give of themselves, both in time and money, to support our family, our daughter, and our cause. I don’t know if people know how touched we are by their generosity. I don’t know if people know how much it means to see Kacy smile, knowing that she has a TEAM behind her for every step of this journey. It is the good that comes from the bad. It is the happy that comes from the sad. It is the hope that comes from despair. Team Kacy raised \$22,000 with this run, but more importantly, we raised awareness and we raised spirits!

“An act of goodness is of itself an act of happiness. No reward coming after the event can compare with the sweet reward that went with it. “

Maurice Maeterlinck



Kacy Wyman and Steve Schleuder

2012 CRN Scholarship for Individuals with Cystinosis

The Cystinosis Research Network has established a scholarship fund to provide supplemental financial assistance to a student diagnosed with Cystinosis who is enrolling in a regionally accredited collegiate or vocational program, or who is currently attending a post – secondary school. The scholarship award, \$1,000, is contingent upon the winner's acceptance to an accredited college, university, or vocational program, or documentation of continued enrollment, and will be payable to the educational institution to be applied toward tuition. An application form is available on the CRN website at www.cystinosis.org/scholarships. For more information or to have an application mailed to you, please contact CRN at 1-866-276-3669 or info@cystinosis.org.

APPLICATION PROCEDURE:

Each applicant must submit:

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

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

Finalists may be interviewed before selections are made.

DEADLINE FOR APPLICATION: Application and all accompanying documents must be received at the Cystinosis Research Network office postmarked **by August 15, 2012**. FAXES OR E-MAILS WILL NOT BE ACCEPTED.

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

MAIL APPLICATION PACKETS TO:

CRN Scholarship Committee
c/o Jessica Jondle
3366 Mt Diablo Blvd 206
Lafayette, CA 94549



Mikaela Gard with one of the cats she takes care of in her pursuit to become a Vet Tech

2012 CRN Sierra Woodward Sibling Scholarship

The Cystinosis Research Network has established a scholarship fund to provide supplemental financial assistance to a student who has a sibling diagnosed with Cystinosis who is enrolling in a regionally accredited collegiate or vocational program, or who is currently attending a post – secondary school. The scholarship award, \$1,000, is contingent upon the winner's acceptance to an accredited college, university, or vocational program, or documentation of continued enrollment, and will be payable to the educational institution to be applied toward tuition.

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

APPLICATION PROCEDURE:

Each applicant must submit:

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

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

Finalists may be interviewed before selections are made.

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MAIL APPLICATION PACKETS TO:

CRN Sibling Scholarship Committee
c/o Marybeth Krummenacker
54 Smith St
Hicksville, NY 11801

Join the Cystinosis Research Network

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

Join The Cystinosis Research Network (CRN) and become part of a global network of caring families, concerned individuals and healthcare professionals working together in the fight against cystinosis. The Cystinosis Research Network's vision is the discovery of improved treatments and ultimately a cure for cystinosis. The Cystinosis Research Network (CRN) is a volunteer, non-profit organization dedicated to advocating and providing financial support for research, providing family assistance and educating the public and medical communities about cystinosis. CRN funds research and programs primarily through donations from the public, grassroots fundraising events and grants.

CRN provides outreach and access to resources. We take great pride in carrying out our motto:

"Dedicated to a Cure. Committed to our Community"...whether you are ...

- A **Parent** who needs critical resource information, support services or help in sharing the challenges of cystinosis to those who serve your child.
- An **Adult** with cystinosis interested in information regarding medical and social issues that are specifically geared for adults.
- A **Relative** or a **Friend** who wants to increase their understanding of cystinosis and find out how you can help out or become involved.
- A **Physician, Social Worker, Educator** or other **Professional** who makes a difference in the life of a family affected by cystinosis, and want to have access to critical information to better serve your patient, student or client.

Joining the Cystinosis Research Network enables you to:

- Receive all the latest cystinosis information through our countless resources, including the biannual CRN Newsletter, our very informative web page www.cystinosis.org, the popular online Cystinosis Support Group, and our toll free number (1-866-276-3669).
- Attend the CRN Family Conference with other cystinosis families to exchange knowledge and create friendships. Also, find out the latest discoveries about cystinosis from the medical professionals.
- Let your voice be heard by legislators and policymakers who need to know why cystinosis (and other rare diseases) are important issues to you.
- Have access to the Cystinosis Research Network's representatives in the areas that are most relevant at any given time to you or your loved one affected by Cystinosis.

Join the Cystinosis Research Network today!

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

Pam Woodward

VP Family Support, Cystinosis Research Network



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.

Please Support CRN's Mission with Your Donation



James Sausman and his cousin, Grace

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

Enclosed is my tax deductible contribution of: \$ _____ made payable to the Cystinosis Research Network (CRN) and mail to: 302 Whytegate Ave., Lake Forest, IL 60045

Name _____

Street _____

City & State _____ Zip Code _____

Phone _____ Fax _____ Email _____

In Honor Of _____

In Memory Of _____

You may send notification of my gift to:

Please check all that apply:

_____ Friend

_____ Individual with Cystinosis

_____ Parent of Child with Cystinosis

_____ Professional

_____ Family

_____ I am interested in volunteering for CRN. Please contact me.

Search the Internet Using GoodSearch and Raise Money for CRN

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

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

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CRN Education and Awareness Update (cont.)

Continued from page 17

My goal has always been to take care of my health as if I have cystinosis but to live my life as if I don't. Leading a "normal" adult life means juggling daily responsibilities most adults have—career, running a household, volunteering, spouse, parents, kids (mine are furry), etc. And there is never enough time to do it all. But I wouldn't trade the hustle and bustle of daily life for a life of wallowing in self pity. There was a time in my life when I was afraid to dream of a "normal" life. I wouldn't buy Christmas wrapping paper at an after Christmas sale because I wasn't sure if I would be around the following year to use it. I didn't put money in my 401K because money was scarce, and I was certain I wouldn't be around to retire anyway.

I have received kidney transplants from four different living donors since the age of 17—from my mom, brother, husband, and friend. I have always been compliant with my transplant medications, and each time my kidney(s) rejected or failed was for a different reason. Now I am learning that if my native kidneys would have been removed, I may have experienced greater longevity with my first transplant. My kidney donors not only saved my life but are role models for unselfishness.

As you can imagine, I have many transplants stories I could tell...from my brother having his donor testing while serving in the Gulf War to my husband's kidney not working for two weeks after it was transplanted. After seven years, my husband's kidney failed from sirolimus toxicity (an anti-rejection drug that can damage the kidney over time). My husband used his credibility as a donor to round up a list of 22 people interested in donating—I never would have been comfortable asking on my own. My nephew and brother-in-law were the best matches, but doctors wanted better. I was on dialysis and working full time, and it was not going well. So it was welcome news when I learned a generous friend from work had been tested and was an 85% match and had no antibodies built up against me. She is the kind of person that when people found out she was donating, they were not surprised. I received her kidney in October, 2009 and have been doing fantastic since. During this time, a doctor in the cystinosis community was my lifeline for advice via cyberspace. She shared a name with my kidney donor, and I will be forever grateful to both of them.

I have been the recipient of so much generosity and miracles from a large team of family, friends, and medical professionals. It is no wonder that sometime during my journey with cystinosis, I found hope and a desire to pay it forward. Now I am not only shopping after Christmas sales, I have become so much of a hoarder that I have had to give some of it away. And I am not only saving money in my 401K but am attending retirement seminars determined to cap off my "normal" life with a "normal" retirement that includes travel. And if I do not get to enjoy my 401K someday, I will make sure that a deserving person or persons benefit from it.

So on my continued quest for a "normal" life, I must bid adieu to the CRN board for the time being. I will still be a part of the community—posting on the message boards and facebook, fundraising, and attending conferences. I am happy to answer any questions, especially about transplantation or muscle weakness, and I may know a living kidney donor or two or four if anyone is considering donating and has questions. Another thing I have learned from my experiences with cystinosis is that life is too short. I need to regroup, spend more time with family, get more exercise, possibly return to school or just take a class or two, finish some projects around the house, and yes, I will still be working. After a couple years hiatus from the board, I plan to come back re-energized, refocused, and ready to help the Cystinosis Research Network again in any capacity I am needed.

T-Shirt Sales Raise Money for CRN (cont.)

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and Whitney Glaize helped gather names for the shirts and helped the process run smoothly. Sue Goulsbra assisted with selling the shirts in the UK.

The shirts were sold for \$20 (adult) and \$15 (child) each, and a portion of the proceeds will go to CRN and other cystinosis organizations. Although the shirts are no longer available for sale, they may be available again later this year. Check the facebook page "Hope has many names" for updates.

Micke had the idea of selling the shirts so that new families would see all the names and know they are not alone. She wants people from all around the world to feel connected and feel hope. Micke is working to establish Hope Has Many Names as a foundation soon.



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Cystinosis is a rare, genetic metabolic disease that causes the amino acid cystine 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. As treatments have improved, children can now live into adulthood and live productive lives.

CRN Vision and Mission

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

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