2017 CRN Family Conference Set for July 13-15 at Snowbird, Utah

The Cystinosis Research Network is pleased to announce that its 2017 Family Conference will be held July 13 – 15 at the Snowbird Ski and Summer Resort located in Utah. Our theme this year is “Soaring to New Heights”.

Plan now to join us and learn about the latest updates in Cystinosis research through lectures, workshops, poster sessions, and medical and patient panels. There will also be opportunities to socialize with other patients and families, which will provide valuable connections and friendships that will last a lifetime. This is a great chance to meet others who are walking in your shoes and also renew prior friendships. CRN is committed to providing family support to those living with Cystinosis. The family conference brings families and the medical community together to share hope and support.

Located up Little Cottonwood Canyon in the beautiful Wasatch Mountains the resort has something for everyone. The lodging will include the Cliff Lodge which is a hotel and also the Lodge at Snowbird which are condos equipped with kitchens. There are lots of activities including Mountain Coaster, Alpine Slide, Mountain Flyer, Vertical Drop, Ropes Course, Climbing Wall, Bungee

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Successful Advocacy Is A+B=C
The President’s Letter from Jeff Larimore

The release of the *The Cystinosis Advocate* about every six months presents the dynamic strides of progress made by the community in so many areas. Over the last few years, the Cystinosis Research Network has successfully expanded on each of its core visions – funding of clinical research grants that has individuals with Cystinosis, their caretakers and extended family cooperating with researchers in continued pursuit of a cure. Secondly, broadening the methods of communication within the community and sharing feedback regarding the effects of current medical treatments and spearheading the possibilities of future improvements. If you consider Research as (A) and Improved Treatments as (B), than the sum creates (C) – Enhancing the Quality of Lives for those with Cystinosis. The stories and recaps that will be presented in this edition of the newsletter will highlight a little bit about (A), a little bit about (B) and a whole lot about (C).

During the calendar year, CRN continued to press the need for legislative support from our national representatives in support of the 21st Century Cures legislation and expansion of membership in the Rare Disease Congressional Caucus. CRN provided patient advocacy representation and presented education material on Cystinosis at the American Society of Pediatric Nephrology, American Society of Nephrology and the World Conference for Lysosomal Disorders medical conferences. CRN was also a primary sponsor for the International Pediatric Nephrology Association medical conference held in Iguacu, Brazil and the Medical Conference for Cystinosis held in Mexico City.

CRN will continue to focus on Strategic Imperatives set forth in 2016. We will continue our dialogues and evaluate platforms to provide a stronger advocacy organization for Cystinosis in the areas of:

- Resources for the Newly Diagnosed through First Decade
- Early Teen Years – Socialization and Transition Education
- Late Teen to Young Adults – Transition Education & Medical Self-Management
- Adults – Defining New Priorities

CRN will continue to press upon medical professional organizations that improving Cystinosis health care education and medical care for all those with Cystinosis is imperative. There remains far too many stories of individuals with Cystinosis and their families feeling stone-walled and nowhere to turn.

CRN is making progress in the area of Global Advocacy Outreach by developing new relationships and exchanging feedback of activities in progress. CRN will continue relationship building with international cystinosis advocacy organizations and representatives in an effort to develop a “one world” list of priorities.

Progressive steps have been made in 2016 for one of my personal goals envisioned in 2014 - “to chart the path of achievement for CRN’s future, a new era needs to begin
where the mission, vision and beliefs...are delivered by individuals with Cystinosis...” I hope you are all excited about the future of the Cystinosis Research Network as I am.

Each Board Member and Committee Member for the Cystinosis Research Network feels deeply in their hearts that their involvement is for a reason and has purpose. CRN always needs to have more families involved. Pat Riley was quoted as saying that “if all members of the team would increase their output by at least 1% more than they had previously over the course of a season, they would achieve a career best”. A little time commitment will blossom into greater success for all.

Continued thanks to Terri Schleuder and to all who submitted articles and updates. To everyone that continues to support the Cystinosis Research Network, our endless gratitude.

On behalf of the Board of Directors and Committees of CRN, good health and happiness this holiday season.

Jeff Larimore
President

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Family Conference continued from page 1

Trampoline, Aerial Tram, Peruvian Chairlift, Gemstone mining and the Cliff spa services. Getting to the Resort is easy, the Salt Lake International airport is only 30 minutes away. For more information about the resort please visit their website at www.snowbird.com

Registration materials will be available in early 2017 on the CRN website and will be mailed to families. Special room rates have been negotiated and will be made available to conference attendees.

We look forward to seeing you in Snowbird in July 2017!
CRN Announces Three 2016 Academic Scholarship Recipients

By Terri Schleuder

This year the Cystinosis Research Network scholarship committee was pleased to be able to award three $1,000 academic scholarships to three outstanding young adults as they continue to pursue their college career goals.

We awarded two Individual with Cystinosis Academic Scholarships, one to Shea Hammond and another to William Patterson. Shea began an MBA Program this fall at Boston College, Carroll School of Management. Over the last 28 years, Shea, has faced many difficult challenges in living life with Cystinosis, including numerous surgeries and three Kidney transplants, but he has never let it stop him from living his life his way. He has always been a compassionate leader, an inspiration and a role model for all of us in the Cystinosis community.

William Patterson with his nephew, James Wyatt

William Patterson is our second Individual with Cystinosis scholarship recipient. Will has also faced many challenges of living with Cystinosis through the years, but has never let it defeat him. Will is completing his BA degree in Journalism and Media Communication at Colorado State University. He plans to participate in a media internship next summer and study abroad in the spring to gain a more worldly view before graduating.

In his own words.... “I am so excited to receive this year’s Cystinosis Academic Scholarship. I am studying journalism and media at Colorado State University in Fort Collins. I intend to use the
scholarship to help me pay for my next semester abroad in Europe and to help support me doing an internship to gain experience in documentary film production."

The last scholarship awarded this year was the Sierra Woodward Academic Scholarship for a sibling of an individual with Cystinosis. The $1,000 award went to Alex Greeley. Alex knows well what living with Cystinosis entails. She has watched her younger brother Jack live with the many challenges life with Cystinosis brings all of his life. Alex is continuing her studies at the Savannah College of Art and Design (SCAD) pursuing a career in Fashion Marketing and Management.

We send our sincerest congratulations and best wishes to all three of these fine young adults as they pursue their life and academic goals.

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**Livgracefully Raises Funds for Cystinosis and Fibromuscular Dysplasia**

**By Kirsten Stilke**

When Livia suddenly left this world, our hearts broke into a million pieces.

Everything we ever knew changed, and we knew that we were never going to be the same. We also knew that within that pain, we needed to find strength to keep pushing forward, for Mason’s sake. In our grief, and with the help of our family, we embarked upon starting something to not only honor Livia, but to help keep her memory alive, in doing what she so loved to do! Thus, "Livgracefully" was born! (Liv's first and middle name together).

We knew that Livgracefully would help us focus on making our little girl proud while doing something amazing. We decided as a family to fundraise and raise money for research not only for Cystinosis but also for Fibromuscular...
Dysplasia (aka FMD); the disease that took Livia from us without an ounce of notice. You see, the kind of FMD that Livia had, could not be diagnosed until it was too late.

So all along, this brave little girl with hair like an angel, and a personality like no other; was fighting not only Cystinosis...but her little body could only fight against FMD for so long before she couldn’t hold on anymore. Had there been more research, perhaps she would still be here with us...

We worked so hard for an entire year to promote Livgracefully and spread the word about Cystinosis and FMD to help us raise funds, so we could lovingly donate to these organizations in hopes for a cure someday! We battle Cystinosis to this day, with our son, Mason who still bravely fights each and every day. But now, he fights this alone. No partner in crime. No sister to hold his hand when he got scared. No one to fully and completely understand him like she did. Even though Cystinosis didn’t take Livia, we still need a cure...we need a cure for Mason and for all the other children and adults fighting this battle each day.

Our loving and generous family donated the first batch of bracelets to us... giving us an awesome head start! For over a year we sold bracelets with the “Livgracefully” logo on them and we were thrilled with the outcome of supporters! We advertised on FaceBook and our website livgracefully.org! We also started a GoFundMe page entitled: Livgracefully as well, which really helped!! It means so much to us that we got the incredibly generous support that we did! We asked a mere $5 per bracelet and all those proceeds went to both CRN and FMD!

We made it our mission to collect until Livia’s 2nd angelversary. (September 26th) We decided that was the day that we would commemorate and honor her
memory by writing out those checks and giving to those entities that are near and dear to our heart! It also
gave us something positive to focus on that day.

For our first ever go at this, we raised a little over
$4,000!!!!!! We split it down the middle and donated
$2,000 each to CRN and FMDSA—in loving memory of
Livia Grace Stilke!

We are already planning next year’s bracelet design and
hoping to add more exciting ideas for fundraising to our
causes! We hope each year we can give back to CRN,
FMDSA as well as starting up scholarships for area
children!

Livgracefully doesn’t take our pain away, but it does help
us focus on something positive. If Livia’s passing
teaches us anything, it has taught us how absolutely
precious life is. How we take for granted the little
moments we should cherish. We are motivated by
making Livia proud, by choosing to Livgracefully; just as
she did in her mere 8 years here on earth. Our hope
someday, is to make Livgracefully an organization that
enriches lives, and helps anything it touches. We know Liv would be proud. Thank you to all of you who
effortlessly and generously supported us in our mission. We could never do it without any of you, so we humbly
thank you with all our hearts!

With Love,
Dave, Kirsten and Mason Stilke
The debut of this section of our newsletter saddens us because of the fact it is needed. It reminds us that even though much progress has been made in understanding and treating Cystinosis over the last three decades we are still far away from a cure. Too many young people still die from the complications of this disease. With each loss we in the Cystinosis community all feel a piece of our hearts die too.

This past August, in less than a week, two more young adults were taken from us, Kwakhe Msibi, age 20 and Weston Tschannen, age 23. Their families have graciously offered to share a bit of their stories with us. One of the lessons Cystinosis teaches us is the beauty and quality of a life is not measured in years, but in the number of lives it touches. The beauty and impact of these two lives is immense. The Cystinosis Research Network sends our deepest sympathies to their families.

I am Constance Msibi, the mother of the late Kwakhe Msibi who passed away on the 29th of August due to Cystinosis. He was a very ambitious young man, very intelligent and positive. He was full of life. He loved himself and people around him. He had so many plans for his future. He was about to do a television show on kidney failure awareness.

I have attached the biography he wrote on February 25, 2016.

**Biography of Kwakhe Msibi**

My name is Kwakhe Msibi. I was born on the 21st of February 1996, in a township called Daveyton, Benoni. I’m currently living in Durban with my mother, Constance Msibi and my two beautiful younger sisters, Sizakele & Khanyi.

At the tender age of two, I was diagnosed with Cystinosis. Cystinosis is a very rare metabolic disease that affects 1 in 1,000,000 children in the world. Thus making it difficult for me to get treatment as it is too expensive and not available in our country. The disease has affected my kidneys, heart, bones and eyes, and I’m currently doing hemodialysis at St. Augustine Hospital, Durban, South Africa.

In 2008, my Father suddenly passed away due to cardiac failure. It was difficult for me to cope - my life was never the same. But with the unconditional love & support from my mother, I managed to pull it through.

Tragically, In January 2014, I got very sick and was admitted at Charlotte Maxeke Hospital in Johannesburg. I reached a stage whereby I was in a coma and in the ICU. I stayed in hospital for half a year I could not attend school that year. My state of health was so severe that the doctors told my mother I wasn’t going to make it. Miraculously, with the faith I had and my Mother’s love for me, I recovered from the
coma. I was so sick that I couldn’t walk or talk. For the months that followed, I was in a wheelchair. I also had a terrible bed sore that left me with a big deep scar on my buttocks and it took a long time to heal.

The doctor told my mother that he is discharging me so that I could go and spend time with my family because I only had a few months to live. That didn’t scare me, I never lost hope I had so much faith within myself. I knew it wasn’t my time yet. I still had a lot to achieve and share with the world. I had so much faith in God that if He was able to take me out of the coma, He is going to save me again. I didn’t focus on the few months to live that was based on science, but I focused on the promise that God gave to me, “I will never leave nor forsake you.” These were the words that my Mother, my rock and my inspiration whispered into my ear everyday while I was in the coma - those words kept me alive.

After my discharge from hospital, my mother took me to hospital for treatment by putting me on her back as she didn’t have a car. Those were the toughest days for my family. Within a few months I had fully recovered.

In April 2015, I went back to school to do my matric. The teachers were hesitant to admit me as I already missed the whole school term. But I assured them that I’m prepared to work hard and complete all the work I missed. I could only attend classes 3 times per week as I had to go to hospital 3 times per week for treatment. That didn’t stop me from doing my school work. I would do my assignments and studies in the hospital bed. My teachers were so proud of my hard work. I completed and passed my Matric in 6 months. I always had this perception that nothing is impossible if you put all your efforts in it.

In February 2015, I was interviewed on a show called Cutting Edge, SABC 1, so that I could talk about my illness and raise awareness to inform the people of this deadly kidney disease. I wanted people to know and understand Cystinosis, kidney failure and organ transplants, and the strength it takes to live with such an illness. It is by no means for the fainthearted or less spirited.

Soon after the broadcast of Cutting edge, a young man came forward and offered to donate a kidney for me. I was so overwhelmed with joy that God is really keeping His promises to me - I am expecting the transplant by June.

Why do I want to do a documentary?

I wish to share my story with the world.

I wish to share my inspiring story with others especially the youth. To encourage them that life is worth living. In order to give others hope, motivate and inspire them to be whatever they want to be regardless of their circumstance or challenges. Some have given up hope, some have turned to drugs, some have committed suicide and some are waiting to die. I want them to know that the challenges in our lives are there to strengthen us; they are not there to run us over.

I have been through many challenges in my life that were life threatening, but I told myself that I wouldn’t let those challenges dominate or take over my life. I have my own inspiration and people who motivate me to be who I was meant to be. Because God has a plan for us all, and it is not to be a failure. I believe and stand by these quotes: “If you can’t get a miracle, become one, and don’t let your challenges determine your future”.

Kwakhe Msibi
The challenges that they are facing now, let them be a stepping stone to their success. They should not put their lives on hold and dwell on the unfairness of past.

IF I BEAT DEATH AND PASSED MY MATRIC AGAINST ALL ODDS WHILE STILL AIMING HIGH...THEY CAN BEAT ANY CHALLENGE AND ACHIEVE THEIR DREAMS AND AMBITIONS.

NOTHING IS IMPOSSIBLE IF YOU BELIEVE AND PUT ALL YOUR EFFORT TO MAKING IT A REALITY.

Weston Tschannen

Weston was born on January 21, 1993 along with his twin brother William. He was diagnosed with Cystinosis at the age of 17 months and took on this disease with a fighting spirit. He never let Cystinosis define who he was. It was just a daily annoyance that he had to endure in order to enjoy all that life had to offer him. A lover of sports, cooking, and music, Weston demonstrated his strength of character by finishing fifth in the 103 pound division of the Missouri State High School Wrestling Championships in 2009. Winning the last match of his 10 year commitment to the sport, Weston defeated his opponent with only 10% kidney function.

Weston suffered complete renal failure at age 16. Receiving the loving gift of a kidney from his mother in 2009, Weston experienced a new lease on life until that kidney failed in 2014, when he began dialysis for the remainder of his life.

Weston further exemplified his determination to overcome life’s challenges by completing his last year of college while undergoing daily dialysis treatments. A year ago, Weston started experiencing Grand Mal seizures and would later be diagnosed with Epilepsy. This was particularly challenging because he could no longer drive. After further testing, Weston was put on two different seizure meds this past March and had not had a seizure since. He graduated in May from the University of Missouri in Columbia with a Bachelor of Science degree with an emphasis in restaurant and hotel management. Most recently, following his
graduation from college, Weston began interning with the research department of Raptor Pharmaceuticals, assisting in the development of programs and town hall meetings to provide help for sufferers of chronic illness.

Weston had a wonderful spirit and such a zest for life! He accomplished so much in his short 23 years and we are so proud of him and so blessed to call him our son. Unfortunately, Weston’s life was cut short on August 28, 2016 due to a very high potassium level and another Grand Mal seizure. His twin brother William, who is an EMT, did everything he could to save his brother’s life but God had other plans for Weston. Devoted to his family, Weston was a loving, funny, loyal companion to his many friends and members of his north Missouri community. Weston will be terribly missed by our family which also includes his brother Ryan and wife Anne, his sister Ally and her fiancé Andrew, and by the many people he touched and those who fought by his side. He taught us so much throughout the years in dealing with Cystinosis and continues to teach us how to live life to the fullest and enjoy every minute!

Loving parents,
Barb & Terry Tschannen

Editors note: This issue begins a new section in our Cystinosis Advocate Newsletter. We would like to invite anyone who has lost a loved one to Cystinosis in the last two years who would like them remembered in our In Memoriam section to submit pictures and a brief story to tschleuder@cystinosis.org. We will include those pictures and stories in future issues.
The Gordon Research Seminar on Lysosomal Diseases is a unique forum for graduate students, post-docs, and other scientists with comparable levels of experience and education to present and exchange new data and cutting edge ideas. The focus of this meeting is to review innovative discoveries in the lysosomal diseases field. The meeting will cover different research areas spanning from basic lysosome biology to the pathogenic mechanisms of lysosomal diseases and new therapeutic strategies for the treatment of these disorders.

After the great successes of the previous Gordon Research Conferences (GRC) on Lysosomal Diseases, the fourth GRC will be held in Tuscany, Italy, March 5-10, 2017. The Gordon Research Conferences provide an international forum for the discussion of the latest scientific discoveries across a variety of fields, and for over 80 years, have been regarded as the world’s premier scientific meetings. The goal of the GRC is to bring together outstanding scientists from academia, industry, and government, ranging from senior principal investigators to Ph.D. students, in a collaborative setting that encourages the sharing of new ideas and unpublished research. We are indeed fortunate to have a GRC focused on lysosomal diseases, and we look forward to building on the success of the previous meetings.

We are currently planning the program for the 2017 meeting and have confirmed the attendance of 40 basic and clinical scientists who are experts in the lysosomal disease field. The 2017 Lysosomal Disease GRC will again offer a critical venue for addressing major topics in lysosomal biology, disease mechanisms, diagnosis and therapy, including (i) lysosomal positioning and movement, which is emerging as a critical determinant of many lysosomal functions: (ii) lysosomal membrane proteins that contribute to cell dysfunction across many disorders and pathogenic cascades (iii) mechanisms of lysosomal storage disorders (LSDs); (iv) lysosomal dysfunction in diseases other than LSDs; (v) new, innovative therapies for correction of lysosomal disease; (vi) diagnostics ad biomarkers for LSDs; and (vii) ongoing and upcoming clinical trials focused on lysosomal disease. A major goal will be facilitation of attendance at this meeting by the most senior investigators in the field, along with junior investigators, postdoctoral fellows and graduate students who are at the beginning of their careers.

Our 2017 meeting has again been selected to host a GRS a special meeting organized and run by graduate students and early postdoctoral fellows working in lysosomal disease with comparable levels of experience to present and discuss cutting edge and
unpublished research. The GRS will take place on the 4th and 5th of March 2017, immediately preceding the GRC. The 2017 GRS is chaired by Chiara di Malta, who works in the chair’s laboratory at the Telethon Institute of Genetics and Medicine (TIGEM), and Ian Martin Williams, ex-postdoctoral fellow at NIH and currently medical writer at Oxford PharmaGenesis UK, who will serve as GRS Co-Chair. All attendees of the GRS are expected to present their work via talks or posters. We aim to have at least 50 GRS attendees, with the vast majority of these individuals going on to attend the GRC.

CRN is proud to provide sponsorship to enable these graduate students and postdoctoral fellows to attend the meeting. This support is in line with one of our organizational goals of providing support to new investigators and clinicians in the field.


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**CRN Sponsored 17th Congress of the International Pediatric Nephrology Association (IPNA 2016)**

*By Christy Greeley*

The Cystinosis Research Network is pleased to announce its sponsorship of the 17th Congress of the International Pediatric Nephrology Association, IPNA 2016, which took place September 20-24 2016, in Iguacu, Brazil.

[www.ipna2016.com](http://www.ipna2016.com) The International Pediatric Nephrology Association (IPNA) is comprised of roughly 1,500 pediatric nephrologists and allied professionals representing 89 countries around the world. Together, they work to disseminate knowledge about kidney disease in children in the areas where care is most needed. Every 3 years, IPNA organizes an International Congress reflecting what is going on in Pediatric Nephrology in the world and presenting the best work of all subfields. Specifically, CRN supported the Cystinosis symposium which will took place during the 2 day pre-congress course on rare diseases and was chaired by Dr. Levchenko and Dr. Nesterova. There was also a congress presentation by Dr. Cherqui on new approaches to Cystinosis during the Renal Tubular Acidosis Symposium.
By Liza McCalla

Kevin and I met in college and I spent practically all my time at his place since it was so much closer to campus than where I lived. We spent ages talking and getting to know each other. We talked about favorite movies, we talked about childhood memories and favorite foods and family members and pets and everything else under the sun.

A few months in, Kevin said that he had something really important to tell me because it was a large part of who he was as a person. I braced myself for the worst and collapsed laughing when the very important thing was that he was spending a lot of time playing a MMORPG (a massive multiplayer online roleplaying game). Only after this did he casually mention one day that he has this disease called Cystinosis and maybe could I help him remember to take his medicine.

I don’t really remember how I learned all the details. I think it was a combination of my researching Cystinosis online, Kevin telling me what he knew and me talking to Kevin’s mom. I knew there would eventually have to be a transplant but it seemed abstract and far away. I wasn’t too worried. I started going with Kevin to his doctor’s appointments once we moved in together. We got married in January of 2007 and it was less than a year into our marriage when Kevin’s nephrologist told us the creatinine was now at
a point where he could be listed for transplantation. Kevin's dad donated a kidney to him in May of 2008.

That kidney was damaged by BK virus after about 6 months. We saw the creatinine steadily creeping up, but in spite of many, many hospitalizations and visits to the ER, nothing really seemed to improve. We got to know the nurses on the kidney floor at our local hospital on a first name basis. The summer of 2010 we moved and as soon as we were in our new place, it became evident that Kevin was having a rejection episode. We got to learn about plasmapheresis and emergency hemodialysis and we got to meet a whole new set of nurses in the process. In October of 2010, the graft was removed due to ongoing pain and Kevin was on peritoneal dialysis at home. We were listed of course, and told that he was automatically at the top of the list and we shouldn't have to wait long.

Kevin says he doesn't really remember a lot of the next 4 years. He had frequent issues with the dialysis catheter flipping internally, affecting the absorption of the dialysis fluid. Multiple abdominal surgeries led to him having internal scar tissue adhesions which caused pain. Constant hospitalizations meant we both missed massive amounts of work and racked up medical bills. We started to play a game to see if we could stay in every room on the kidney floor at our local hospital. After being in the hospital one Thanksgiving, we started keeping track of which holidays we were in the hospital too. As it stands, we stayed in all but about 9 out of 36 rooms on the floor, and were in the hospital for both of our birthdays, our anniversary, and all the major holidays except for Christmas and New Year's.

Kevin got admitted on Jan 2nd one year and we missed the chance to check off New Year's. (I should stress here that this was really my game more than his, since he was usually medicated.)

Throughout it all, I learned that Kevin is not a good historian when it comes to his medical history. Kevin
The Cystinosis Advocate

Kevin and Liza welcome nephew, Samuel Patrick Salzar.

also does not like to push and ask more questions and insist. Luckily for all of us, I have no qualms about asking more and insisting if I feel it's necessary. I made it my business to be knowledgeable about every aspect of the disease and every new procedure that was being done. Imagine my surprise when Kevin did not want to see the pictures one surgeon took for me of internal scar tissue adhesions. (They are fascinating looking, like fleshy stalagmites). Kevin learned that he can't have children because I read a study that showed that males with Cystinosis did not have viable sperm. We later confirmed this with Dr Gahl at NIH.

While on dialysis, I "diagnosed" him with Restless Leg syndrome and sleep apnea, with actual doctors later confirming both my assessments as correct. Asking Kevin if he's taken his medicine is such an ingrained habit by now that I usually ask him 5 or 6 times even after he's answered me that yes, he has taken it.

This may sound overwhelming and it occasionally is. Being an adult with Cystinosis has a completely different set of challenges than being a child with Cystinosis. Being somebody married to an adult with Cystinosis is something else entirely. It can feel overwhelming and unfair. Spouses can start to feel like caregivers and get caregiver burn out. For a split second, you'll wonder if your relationship is really worth it when
you spend hours on the phone with Medicare, trying to work out why they are refusing to pay. While I have always admired people who say that having the disease is a blessing, or people who feel that it’s a small burden to bear in comparison with diseases others may have, I have not found this attitude helpful for myself and Kevin. The idea that you should not be happy with your lot because somebody else has it worse is as strange as if you believed you should be unhappy with your lot because somebody else is more fortunate. We are allowed to feel overwhelmed and be sad that Kevin’s day to day reality includes 48 pills. Kevin is allowed to be sad that his body always requires such constant, careful treatment in comparison to other men his age. These things do not make us weak, they make us pragmatists.

It’s not all doom and gloom though! 2 years ago, after 4 years of dialysis, Kevin’s sister donated a kidney and through a paired donation involving 3 sets of donors and recipients, Kevin now has a new kidney that is working well. His overall health and wellbeing has improved tremendously. He got a commission to work on a series of comic books about Cystinosis which allows him to get paid for what he always wanted to do while drawing and writing about a subject he’s intimately familiar with.

Every marriage has it’s challenges, and this is manageable for me. I liked Kevin more than I was afraid about his disease. It’s our 10th anniversary this coming January. We may not have children but we have four fantastic cats and a beautiful niece and nephew we get to see frequently. We have the support of Kevin’s family and the wider Cystinosis community. I don’t consider Cystinosis to be a gift or a blessing or something that makes us stronger. I consider it as something we can manage because we are strong.
Cystinosis: a truly orphan disease.
Report of the Cystinosis Foundation India

By Rajan Ravichandran,
Chairman, Cystinosis Foundation India; Director, MIOT Institute of Nephrology, MIOT Hospitals, Chennai, India

(original article from RARE DISEASES AND ORPHAN DRUGS An International Journal of Public Health)

Abstract

Cystinosis is a rare disease leading to accumulation of cystine in lysosomes causing apoptotic cell death leading to organ dysfunction. Although the disease was identified 100 years ago, the history of cystinosis in India is really pathetic. Only in 2012, the Cystinosis Foundation India was formed with the initiative to pool up these unfortunate patients. Nineteen patients have been identified and registered with the foundation. Out of these, only 8 are receiving specific therapy with cysteamine. Four patients have undergone successful kidney transplantation. Eight patients have died since registering with the foundation. Seventeen patients were picked up with advanced growth retardation and renal failure. Only 2 children were picked up in early stage. This article describes the difficulties faced in the identification and management of these patients in India.

Key words

Cystinosis, renal failure, growth retardation, cysteamine, India.

Introduction

Cystinosis is an autosomal recessive disorder caused by mutation of the CTNS gene on chromosome 17 which encodes a ubiquitous cystine specific transporter in the lysosomal membrane [1]. There is massive intra lysosomal accumulation of cystine due to the transport defect leading to apoptotic cell death and progressive organ dysfunction. The disease manifests itself around 6 months of life with failure to grow. Signs of Fanconi’s syndrome (polyuria, polydipsia, dehydration and acidosis) also appear as early as 6 months. Corneal crystals diagnostic of cystinosis can be present before 1 year, but is always present after the age of 16 months [2]. Untreated, the children progressively develop rickets, severe growth retardation, renal failure and die between 10-15 years of age. The treatment of the disease essentially is to identify early in the first year of life, replace the nutrients and specific treatment with cysteamine. The diagnosis is confirmed by estimating the leukocyte cystine level and identify the genetic mutation. In 1976, Crawhall reported the news of cysteamine resulting in efflux of cystine from the lysosomes [3]. Gahl showed that protracted oral therapy with cysteamine depleted the organ cystine and delayed the complication of cystinosis [4]. So far there have been hardly any reports of cystinosis from India. Phadke et al reported in 2004 a 3 year-old child who presented with Fanconi syndrome with mild renal failure and corneal crystals [5]. The child was initiated on treatment and lost follow-up. In 2014, Krishnan
Swaminathan reported the agony of a boy whose diagnosis of cystinosis got delayed in spite of visiting several hospitals ultimately presenting with severe renal failure and growth retardation [6]. The sister of that patient had also died at the age of 7 of a similar condition. In 2015, Sharma reported the biopsy finding in a 3 year old child with cystinosis and renal failure [7]. Akhilesh Kumar and Bachhawat have discussed the molecular basis of cystinosis [8]. Taosheng Huang reported the details of CTNS mutation in an Indian boy with nephropathic cystinosis [9]. The first successful kidney transplantation in a severely growth retarded child with cystinosis was reported from Chennai, India in 2010 [10]. Subsequently, when the child attended the school, he was not able to see the black board and the eye examination confirmed the presence of crystals in the cornea. The diagnosis of cystinosis was made retrospectively and his 2 year old brother was identified with the disease on further investigation. It was this episode that lead to the formation of Cystinosis Foundation of India in 2012. The foundation was launched on May 2012 in Chennai by a NGO Sapiens Health Foundation. Important members of the society from different professions like law, accountancy, journalism etc. were made advisors of the foundation. Donations were raised from the general public to run the foundation.

**Materials and methods**

After the foundation was launched in Chennai, several nephrologists and ophthalmologists in leading centers throughout India were contacted to register their patients with cystinosis. Booklets were distributed in leading nephrology conferences held in the last 3 years. Once the patient was registered, the clinical details including the biochemical workup were included in the records of the registry. Approximately 1100 nephrologists were contacted throughout the country by email correspondence. This paper highlights the clinical data of these patients, the lack of treatment for cystinosis throughout the country as a whole and the difficulties faced in procuring the drug for these patients.

**Results and discussion**

The effort of the foundation bore fruit and 19 patients have been registered so far in the foundation. Out of the 19 children registered, 4 are females and rest males (Table 1). Only 4 children could be picked up below the age of two. All the children had growth retardation and Fanconi syndrome (Figure 1). Only in 2 children, the creatinine clearance was normal. Seventeen of them had renal impairment including 4 patients having undergone kidney transplantation. One patient continues to be on peritoneal dialysis after failed graft 12 years ago. She is the eldest with the age of 20 years. 8 patients have already died after registering in the foundation. Only 8 patients are continuing on specific treatment with cysteamine although 13 were initiated. None of the patients are using the cysteamine eye drops. Consanguinity in the parents was noted in 11. Thirteen patients had hypothyroidism. Corneal crystals were found in all the patients. Three of the children had an affected sibling. Three other children gave a history of similar illness in the sibling who had died earlier. In 2 of the children, the disease was picked up early in life because of the correct diagnosis in the affected sibling. The treating physicians were contacted, giving more information about the disease and the need for specific treatment. The test, cystine estimation in leukocyte, is not available in India. Hence samples have to be sent to USA for confirmation. Next came the biggest challenge procuring cysteamine for the patients. Cysteamine is not approved for use in India. Hence Orphan Europe could not sell the product in India. The individual patients have to apply to the drug controller for a special permission to import the drug. After that, money had to be remitted to Orphan Europe by individual patients. The cost of the drug is phenomenally expensive with none of the patients getting reimbursement from insurance. Approximately 200 euros is required for a treatment period of 3 months per child. The drug which is then sent by courier requires to be cleared from customs which again involves a 5 to 20% duty. Out of the 19 patients, only 13 were able to procure the drug. Five patients on the drug have died. At present 8 patients are continuing the drug. The Foundation raises the money by donation and is sponsoring the drug for these patients. Attempt has been made to manufacture the drug locally. The eye
drops which are required to improve the corneal deposit are not available. The government does not permit the drops to be formulated by the pharmacy. Big pharma companies are not interested in manufacturing the eye drops because of the low number of patients. It is almost impossible to import the eye drops since it is much more expensive than the oral drug. Attempts have been made to contact Orphan Europe, Raptor Pharma & Sigma Tau Pharma to apply for approval of the oral drug and eye drops in India. This situation can be dramatically improved if pediatricians pick up the disease in the first year of life itself, so that treatment can be instituted before the child develops complications. This is possible if the test for cystinosis is made available at least in the major cities of India. The drug cysteamine should also be indigenously manufactured, bringing down the cost resulting in adequate therapy to all patients. The Indian Institute of Technology (IIT, Madras), a quasi government organization, has already produced the drug cysteamine in the laboratory and one of the pharmaceutical companies has agreed to manufacture commercially.

**Conclusion**

Thus, cystinosis has been a totally neglected disease in India with very poor awareness amongst the medical fraternity to pick up the disease early. Procuring cysteamine has been very expensive and difficult. The only light seen is the formation of the Cystinosis Foundation, India in 2012 with subsequent attempts in the right direction.
Acknowledgements
The author wants to thank Kannan S, Research Coordinator, Madras Institute of Nephrology Research Pvt. Ltd, Chennai, India

References
10. www.cystinosisindia.org
Angels Among Us

By Katie Larimore

(Editors note: Sometimes stories come to us that beg to be shared, and demonstrate how connected we truly all are. This is one. The piece below was originally shared on Facebook by Katie Larimore, mom to Sarah with Cystinosis and wife to CRN president Jeff Larimore. The military man mentioned, in the post cited a program called HomeBase in Boston with "saving his life" following a difficult readjustment to civilian life after military service. That program was founded by General Jack Hammond, father to Shea Hammond with Cystinosis. Angels truly live among us and intersect our lives at times we need them the most.)

This is probably the most important and beautiful post I have ever written. As my friends know our daughter Sarah has an ultra rare metabolic disease called Cystinosis. Yesterday we were traveling home from a three day trip to the National Institutes of Health for a clinical study. It was a rough travel day as our flight was cancelled and we were diverted to New York ... We finally caught a flight to Charlotte in literally the very back of the plane... Last row by the bathroom! This is actually the best seat for a kid with Cystinosis so we did not complain at all! After two trips to the bathroom and apologizing to the man across the aisle, this obviously military man leaned over and said "no problem, I understand because I have to use the bathroom just as often".

We got to talking and he explained that he was coming home from a three week program in Boston called HomeBase. This is an incredible program for veterans returning from the hell of war who are having trouble with adjusting to civilian life. This man had retired after more than 30 years of service in the Army, most recently at Fort Brag in Fayetteville, North Carolina. He is an Iraq and Afghanistan Vet who is recovering from the emotional and physical damages of war.
He said the HomeBase program has saved his life. He also said his injuries cause many of the same problems that Sarah faces. He spoke with her as a friend and told her he "gets it". I told him about my Daddy who is a Vietnam Vet. He told us about his wait for a service dog to help with his anxiety. As a dog lover I related to his hope for a German Shepherd. As we left the plane he stopped us. He addressed Sarah by name and reached in his pocket. He removed this stone and handed it to her. He said it was given to him upon his return from Afghanistan and had helped him face some scary stuff. He said he wanted Sarah to have it and be strong. When she felt nervous or scared just rub this stone. It had never let him down. Sarah has not let this stone out of her sight. I hugged him in the middle of Charlotte Airport at 10 pm last night. I do not even know his name. There ARE Angels all around us!

Rovithis Realty takes a percent of each sale and donates that money to a charity of the home owners choice. For Jenn Loglisci’s sister and her husband the charity of choice was an easy one. Thank You!!!
Hello Friends,

Our Art exhibit is now available online! Every piece of art and every artist are profiled on our new website. If you have not yet seen the talent our community has put on display please visit www.dreamachieveinspire.org to take a virtual tour of this amazing project!

Also big news…. Our very own comic artist, Kevin McCalla, has released his first comic for our community! Super Cysteamine Team is available to all free of charge! Sign up on the CRN website www.cystinosis.org or the link https://cystinosis.org/news/announcements/225-cystinosis-comic-book-series. Once you sign up the first book will be sent and you will automatically receive the next two stories! Everyone is invited to enjoy the adventure!

I would love to hear your reviews and comments! Klarimore112@sc.rr.com

Dream a little today!
Katie Larimore

Elsie Buck loves reading the Super Cysteamine Comic book

Note: Kevin McCalla is our Featured Adult in this issue. Please read more about Kevin on pages 14-17
Marybeth Krummenacker meets with NORD Founder Abbey Meyers

By Marybeth Krummenacker

Marybeth Krummenacker had an opportunity to spend some time and have lunch this summer with Abbey Meyers. Abbey is the founder of NORD (National Organization for Rare Disorders) which was the driving force behind the Orphan Drug Act of 1983. Without that important piece of legislation, the Orphan Drug community would not be where it is today. The Cystinosis community is so grateful to Abbey for her dedication and hard work to see that this legislation became law! Without the Orphan Drug Act of 1983, the Cystinosis community would have never had a medication developed. Cystagon is #41 on the list of the first 100 drugs approved as a direct result of the Orphan Drug Act!! We in the Cystinosis community are blessed to have been at the right place at the right time. We in the Cystinosis community are so grateful to Abbey for her years of dedication to NORD and Marybeth was very proud of her 6 years served on that Board. The afternoon spent was filled with many wonderful stories and a lot of laughs and Abbey invited Marybeth back again! She is hoping to take her up on that offer soon! If you are interested in reading Abbey’s story, “Orphan Drugs: A Global Crusade” you can purchase it on Amazon. Marybeth said that she thoroughly enjoyed her time spent with Abbey...“it is not often that you get to meet one of your personal heroes...but I did and I am blessed”.

NORD founder and past president, Abbey Meyers has lunch with Marybeth Krummenacker this past summer
José Morales and Marybeth Krummenacker represented Cystinosis Research Network (CRN) at the Global Genes’ Rare Patient Advocacy Summit and Tribute to Champions of Hope. Global Genes is one of the leading rare disease patient advocacy organizations in the world with over 500 global organizations in its membership. This year’s event was held at the Hyatt Regency in Huntington Beach, California. The goal of the Patient Advocacy Summit was for patients, caregivers, and advocates to walk away equipped with actionable steps to increase diagnosis, assist in building self-advocating communities, promote funding of research, engage in developing treatments, therapies and advocating for rare diseases.

Over the course of three days and evenings, over five hundred participants representing patient advocacy organizations, medical and research institutions engaged in a series of plenary and specialized tracks. The theme of the three days of meetings was --- LEARN, CONNECT, INSPIRE. All attendees were encouraged to Learn by not only attending sessions but also engaging the presenters and asking probing questions. The opportunity to Connect with other rare disease advocates and industry experts was encouraged throughout the three days and there was an abundance of opportunities to do just that! The Inspire theme was for the broader community and we were encouraged to share via...
social media channels our impressions, learnings and to look for change within your own communities.

There were three principle tracks promoting the themes of:

“How To”
“Care and Support”
“Patients Role in Drug Discovery”

These sessions were rich in information for those in attendance to select from. Marybeth and José attended a multitude of sessions and extracted valuable insights which will be utilized to further enhance and refine CRN’s own operating model, as appropriate. A highlight of the summit was the opportunity to speak with Dr. Alvin Shih, Retrophin Executive Vice President and Head of Research and Development. Dr. Shih was a key contributor on a panel discussion addressing “Game Changing Science, Technology and Clinical Care - What Patients Should Expect”. The lively discussion featured Ryan Taft, Senior Director of Scientific Research, Illumina, Inc., and Dr. Anthony Philippakis, Chief Data Officer from Broad Institute. There was an energized interaction amongst panelists and the audience about the fast paced world of technology and the lightning speed with which things are changing.

As emphasized throughout the event, a rare diagnosis changes everything. We as parents and caregivers are not given a playbook on how to cope or take the next step or even understand science and medicine. We have certainly not been handed a blueprint on how to build an advocacy organization or successfully bring a promising therapy to market. We learn from each other and the good news is that rare disease advocates are some of the most inspiring on the planet! Global Genes is one of the organizations that works towards bringing the community of “RARE” together to share best practices, create important introductions and help to enable collaboration. CRN continues to support and advocate for the Cystinosis community.
Midwest Cystinosis Gathering Held in Iowa, July 8th-10th, 2016

By Doretta Hoffmann

The Midwest Cystinosis Gathering was held on the weekend of July 8th - 10th with Bill Croce along with the Jay and Khristy Rollinger family hosting the event in Bettendorf, Iowa. We kicked off the weekend with a dinner prepared by the host family in the courtyard at the hotel. It was a fun evening gathering together to catch up on what everyone had been doing over the past year.

Those attending this year’s event were Tresa and Shawn Reuter, Holly and Lonnie Hohl, Sarah, Andrew, Jon, Doretta and Don Hoffman, Candy, Nick and Don Waggoner, Emily Swim, Jay and Khristy Rollinger and Bill Croce.

On Saturday morning we all gathered to go on the Mississippi River taxi ride. This event was a great way to tour the Quad Cities area along the Mississippi River. We were very grateful to have the CRN provide us with a delicious lunch on Saturday. The afternoon was spent visiting and a few people trying their luck at the casino. On Saturday evening we attended the Quad City River Bandits baseball game, which ended with the River Bandits on the losing end but it was a fun evening.

Sunday we had breakfast at the Rollinger family house and we decided that the 2017 Midwest Cystinosis Gathering will be hosed by Don & Candy Wagner family in Grinnell, Iowa with a date to be determined.
American Society of Nephrology Conference 2016 in Chicago

By Rachel DaLomba

CRN attends the 50th annual American Society of Nephrology Conference (ASN) that was held in Chicago, Illinois in November of 2016.

CRN’s Executive Director, Christy Greeley, along with Rachel DaLomba and Anjie Polanco, represented CRN as exhibitors at the ASN 2016 event. CRN provided attending physicians with a plethora of literature, including our new educational comic book, *The Super Cysteamine Team*. It was also great to see many familiar faces at ASN this year and be able to provide them with this creative educational tool. However, CRN had many new physicians seeking information about Cystinosis.

We are grateful to have had the opportunity to continue to raise Cystinosis awareness and celebrate ASN’s 50th anniversary.
Inherited kidney diseases represent a huge burden for the affected patients, mostly children, and their families. The idea of setting an African group for inherited kidney diseases has been met with encouraging response from African pediatric nephrologists in the African Pediatric Nephrology Association (AFPNA). That was crucial to set the **African Inherited Kidney Diseases working group (AfrInKiD)**. With the tremendous advances in diagnosis and management of inherited kidney diseases, it seemed clear more than ever that an African body to care for inherited/rare kidney diseases on a continental level is desperately needed. Since Africa is mostly dim on the World map for rare/inherited diseases, this group should help organizing efforts to improve provided care, build up registries and foster continental as well as international research collaboration into these diseases.

**Why AfrInKiD?**

AfrInKiD was chiefly established to address the deep concern of African pediatric nephrologists regarding the status of inherited kidney diseases across the continent. It is set up as a platform designed to improve provided care and patient-informed practice, ensure access to orphan drugs and foster intracontinental as well as international translational research collaboration into these diseases.

**AfrInKiD Logo**

It is designed to show DNA strand linking the African continent with its five regions (Northern, Eastern, Western, Southern & Central) to the kidney.
Action plan:

The action plan rests on four pillars, setting out immediate actions to tackle:

1. **Awareness, education & training**
2. **Networking & regional referral centers**
3. **Standards of clinical care & registries**
4. **Genetic studies & collaborative research**

**Cystinosis in Africa**

The awareness of Cystinosis is lacking in most of the continent (with very few exceptions) given its rarity that implies limited knowledge of diagnostic tools and therapeutic regimens. Consequently Cystinosis is largely overlooked and the clinical care of affected patients suffers from major problems. Cystinosis awareness & education program is being developed to look closely for Cystinosis across the continent, facilitate diagnosis and set standards for clinical care with guaranteed access to cysteamine therapy (both oral & eye drops).

**Networking & Collaboration**

*AfrInKiD* intends to work closely and network with existing global bodies to develop "African expert referral centers". Moreover fostering international collaboration is pivotal to generate new scientific knowledge into the causes and mechanisms of inherited nephropathies. African contribution to international inherited kidney diseases’ registries is expected to have huge impact on our understanding of these diseases, as shown by the emerging research from few African countries so far.

*AfrInKiD* members with African pediatric nephrologists during the 17th IPNA Congress - September 2016 - Iguacu, Brazil
Finance Update

By Jeff Larimore
The Cystinosis Research Network, Inc
Financial Review – Accrual Basis

For the nine (9) months ended September 30, 2016

Revenues

For the nine months ended September 30, 2016, total income collected of $171,000 was approximately 70% less than the same period in 2016. This decrease was attributable to a reduction in fundraising events in 2016 along with less corporate grants being secured to support program services. Favorably, third quarter 2016 total income was consistent with total income collected during the third quarter of 2015.

Expenses

Total operating expenses of $132,000 were 75% less than operating expenses for the nine months ended September 30, 2015. A decrease in Education and Conference expenditures during 2016 is the source of the expense reduction. Total General & Administrative costs through September 2016 were slightly higher than the same period in 2015 as professional audit costs were billed earlier. G&A costs as a percentage of income increased from 6.0% to 18.0% due to the reduction in 2016 inflows.

CRN has funded $58,000 in research grant installments through September 2016. No research grants had been funded during the nine months ended September 2015.

CRN had net operating loss of $20,000 for the nine months ending September 30, 2016 as compared to net operating income of $30,000 through September 2015. Successful fundraising activities and corporate support in 2015 has carried forward to provide cash resources to increase patient advocacy activities in 2016 to support the Cystinosis community.

Cash on hand at September 30, 2016 was $302,000. Net change in cash for the last nine months is a decrease of $14,000. The decrease approximates the amount of change within the balance sheet accounts through September 2016.
Designate the Cystinosis Research Network as your AmazonSmile charity of choice

Support our organization each time you shop with Amazon. Designate the CRN as your favorite charity and Amazon will donate 0.5% of eligible purchases back to the Cystinosis community.

Simply follow these 4 steps:
- Visit https://smile.amazon.com
- Select Your Account
- Under Settings select Change Your Charity
- Type and select Cystinosis Research Network

Once you are set up, shop https://smile.amazon.com instead of https://www.amazon.com/ and the donations are automatically sent to the CRN. Wishing you happy holidays and happy shopping!

#GivingTuesday is a global, charitable movement observed on the Tuesday following U.S. Thanksgiving and the widely recognized shopping events Black Friday and Cyber Monday.

For the second year in a row, the CRN is an official #GivingTuesday partner. Leading up to and through November 29th, 2016, we encourage you to do a charitable act – whether that involves time, money, or lending your voice to increase Cystinosis awareness.

#GivingTuesday is fueled by the power of social media and collaboration. Please visit our Facebook, Twitter, and Instagram accounts for ordinary ways to be a part of something extraordinary. Last year, 70,000 people across 70 countries came together on this day to raise $116 million. To make a #GivingTuesday donation to the CRN, visit https://cystinosis.org/how-to-help/donate.
Research Update

By Christy Greeley, VP Research, Executive Director

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 funded over $4 million total in research grants and fellowships. CRN has funded a Cystinosis fellowship at the National Institutes of Health and has funded research and education programs in the United States and many countries around the world including Egypt, Mexico, England, Scotland, Italy, Belgium, France, Germany and much more. CRN research topics are aimed at every aspect of Cystinosis with the purpose of understanding the disease and finding improved treatments and a cure. Topics include research and therapies related to neurological, genetic, ophthalmological, gastrointestinal, muscular, nephrology, pulmonary, skin, improved medications, and psychological effects of the disease.

CRN is currently funding three grants, with new projects in the works. CRN will issue another Call for Research Proposals in the spring of 2017. These three grants represent one established Cystinosis researcher in Dr. Levchenko’s group in Belgium, one esteemed neuroscience lab at Montefiore in the Bronx which is expanding their work into Cystinosis, and one innovative newborn screening initiative in Germany led by Dr. Hohenfellner. Following are the latest updates on these projects.

Research Grant Updates

Mechanisms Underlying Neurocognitive Changes in Cystinosis, John Foxe, PhD Co-Principal Investigator, Sophie Molholm, PhD Co-Principal Investigator, Steven U. Walkley, DVM, PhD Co-Principal Investigator

Cystinosis has long been known for its significant impact on renal and thyroid function. It is only in recent years, however, due to the emergence of effective life-prolonging treatment regimens for these primary clinical symptoms that researchers and clinicians have been able to turn greater attention to the impacts this disease has on brain function. To date there is only limited work in this area. To address this shortcoming of knowledge, investigators in Pediatrics and Neuroscience at the Albert Einstein College of Medicine (Molholm, Walkley) and the University of Rochester Medical Center (Foxe) are evaluating individuals with Cystinosis as well as mice in which the Cystinosis gene has been knocked out.

The human studies involve the use of high-density electroencephalographic (EEG) recordings of brain activity being carried out by Drs. Molholm and Foxe in Einstein’s Cognitive Neurophysiology Laboratory. Dr. John Foxe, lead investigator on the human project, reports observing a strikingly "normal" pattern of multisensory behavior and brain responses in Cystinosis, a finding that stands in stark contrast to those obtained in another lysosomal storage disorder where significant impairment is observed (Niemann-Pick type C disease). The group will continue to collect data for this study,
to see if this finding holds up in a larger sample. In addition, following these "positive" results indicating intact sensory processing, they are turning to measurement of cognitive processes that require highly coordinated activity across extensive networks of cortical regions, as these are likely to be more sensitive to any neural damage incurred.

For the mouse studies, spearheaded by Dr. Walkley, director of Einstein’s Sidney Weisner Laboratory of Genetic Neurological Disease, mice with genetically-induced Cystinosis have been established in a breeding colony and are being evaluated for changes in selected brain regions (hippocampus, neocortex and cerebellum) in an attempt to determine with greater precision just how this genetic disease impacts the function of individual types of brain cells. Analyses here range from exploring connectivity and the structure of individual neurons to changes in the metabolic activities secondary to the disease-induced defect in lysosomes. As phenotypic biomarkers related to Cystinosis are identified in these brain regions, the impact of treatment (e.g., with cysteamine) in preventing, delaying and/or reversing these changes will be pursued.

The ultimate goal of these tightly collaborative studies from the two labs is to more fully understand the effects that Cystinosis has on brain structure and function and how factors leading to such compromise could be alleviated.

Altered protein kinase signalling as a cause of reduced adhesion and increased motility of renal epithelial cells in Cystinosis – 6 month report, June 2016, E. Ivanova, L. van den Heuvel, E Levchenko (Principal Investigator)

Cystinosis is a genetic disease manifesting early in life (∼6-12 months) with progressive kidney disease resulting in renal failure early during childhood if not treated. In Cystinosis the metabolism of the amino acid cystine is defective leading to its accumulation in the kidney and other organs. This cystine accumulation results in cellular damage and death, but the direct mechanisms beyond this phenomenon are largely unknown. Some harmful cellular events in Cystinosis might not be directly related to cystine accumulation and are the subject of our research project.

Based on our previous work we hypothesized that the loss of highly specified renal cells like glomerular podocytes and renal proximal tubular cells in urine is a major mechanism causing renal pathology of Cystinosis. Increased rate of cellular abundance in urine can be explained by either the decreased adhesion of renal cells to their matrix or their increased motility or by a combination of both mechanisms. Indeed we demonstrated that both events
occur in cultured human renal cells derived from Cystinosis patients.

We further tried to explore the mechanisms beyond this cellular loss. It has been reported in other diseases that increased cell motility and defective adhesion can be associated with the altered protein kinase signaling. In cystinotic podocytes we found an increased expression of activated or phosphorylated Akt kinases compared to control cells. This could explain, at least partially, the abnormal phenotype. We are currently testing other protein kinases that might contribute to this mechanism. In addition we tested the gene expression of several integrin in podocytes, as podocytes adhere to the extracellular matrix using integrin receptors. Although only minor differences were found between cystinotic and control cells, cell surface expression of these proteins still has to be studied. So far most of our experiments were done in podocytes. We recently started to investigate proximal tubular epithelial cells which also showed an increased expression of phosphorylated Akt kinases unifying the concept of the hypothesis over different renal cell types. Our future plan includes also the experimentation with different kinase inhibitors to explore if they can reverse abnormal renal phenotype.

**Symposium on bones and muscles in Salzburg, Austria, December 8-10, 2016**

Christy Greeley will be attending this conference as an invited patient representative on behalf of CRN and the Cystinosis community. The conference is planned and hosted by Dr. Katharina Hohenfellner, Cystinose-Zentrum Traunstein, Germany. Dr. Hohenfellner is a CRN funded investigator. Her group is working on postpartum screening for Cystinosis with urine dipsticks in 12 week old infants, among other Cystinosis projects.

The goal of the meeting is to set international guidelines for preventing and treating bone and muscle problems in Cystinosis patients. This meeting is meant to find solutions, therefore representative clinicians and researchers for every discipline connected with Cystinosis have been invited -- all in all 21 specialists from many countries including specialists in osteology, physiology, laboratory medicine and pharmacology.
DO YOU OR SOMEONE YOU LOVE HAVE CYSTINOSIS?

Are they affected by:
Photophobia?
Eye Pain?
Foreign Body Sensation?
Squinting?

If you said “yes” to any of the above, it may be time to discuss these symptoms with your ophthalmologist.

Cystinosis causes cystine build-up in the body which may damage cells in the kidneys, liver, brain, other organs and the corneas.
Education & Awareness Update

By Terri Schleuder – Vice President of Education & Awareness

As the year 2016 draws to a close, CRN is beginning extensive preparations and planning for the upcoming Family Conference to be held in Snowbird, Utah July 13th -15th ,2017. Our theme this year is “Soaring to New Heights”. Over the last year we have endeavored to do just that as we continue to provide support to the community, educate and raise awareness about Cystinosis and provide financial support for research offering promising treatments to increase the quality of life for all who live with this disease every day.

We were able to offer three $1,000 academic scholarships this year and congratulate the very qualified recipients as they continue to pursue their collegiate goals. Shea Hammond and Will Patterson each received an Academic Scholarship for an Individual with Cystinosis. Alex Greeley received the Sierra Woodward Sibling Academic Scholarship offered to the sibling of an Individual with Cystinosis. (read more about them on pages 4 and 5 of this newsletter).

There were many Town Halls and regional family meetings held over the year in Austin, Portland, Boston, St. Louis, Indianapolis, and Pittsburg that offered opportunities for families to connect in small groups, ask questions and learn more from experts in attendance.

CRN was represented at the Global Genes Rare Patient Advocacy Summit held in Huntington Beach, California, from September 22nd-24th by Marybeth Krummenacker and José Morales. (Read more on page 26-27)

In late October, a Cystinosis Adult Summit was held in Charlotte, NC. Hosted by Aimee Aldemann, it provided an opportunity for adults with Cystinosis to connect, discuss and learn from each other about their unique experiences and plan for the future.

We exhibited at the Pediatric Academic Society (PAS) meeting held in Baltimore in May and at the American Society of Nephrology (ASN) Conference held in November in Chicago this year. The opportunity to raise Cystinosis awareness among the many attending physicians is highpoint in our year.

Earlier in the year we sent several members of the Cystinosis community to Washington...
Rare Disease Day 2017 Theme Announced

The tenth annual Rare Disease Day will be celebrated on February 28, 2017. The theme this year focuses on Research. Research brings hope to people living with a rare disease. Hope for answers, solutions, better treatments, and ultimately a cure. Learn more about this most important day in the Rare Disease Community and how you can become involved by clicking on the link below.

"Rare Disease Day 2017 is therefore an opportunity to call upon researchers, universities, students, companies, policy makers and clinicians to do more research and to make them aware of the importance of research for the rare disease community."

http://www.rarediseaseday.org/article/get-involved
CRN Development Update

By José T. Morales, Vice President of Development

I have to admit I have had my hands full with the ebb and flow of life these last few months. Just when I thought I was getting my hands around the tasks at hand, another requirement or new variable emerges. I expect this scenario is familiar to all of you. Mind you, I am not complaining. The alternative is significantly less desirable. One of the tasks at hand is drafting my 2016 Fall/Winter Development Newsletter article. I started to write it multiple times only to be distracted or diverted to other activities or interests. So, today is the day! There isn’t any time left for me to entertain myself by reading the latest breaking news about the presidential candidates. Better not go there! The leaves started to change color a few weeks back and are now beginning to fall here in Connecticut. The temperatures have dropped and we have brought out our fall and winter clothing. The Thanksgiving and Christmas holiday season will be here before you know it. Exciting times lie ahead.

We had had exciting times for our Cystinosis community as well. We continue to forge ahead with our three principle pillars:

**Research**
- Responsible for identifying and prioritizing research activities
- Responsible for managing:
  - Professional Advisory Committee
  - Medical Advisory Committee
  - Scientific Review Board
- Responsible for identifying and requesting grant applications from researchers interested in conducting research projects that will lead to improved treatments and / or a cure for Cystinosis
- Responsible for reviewing grant proposals with the Scientific Review Board
- Responsible for assisting with organization of the biennial Family Conference

Christian and José Morales enjoy the Global Genes, Rare Patient Advocacy Summit & Tribute to the Champions of Hope Gala, held in September in Huntington Beach, California
Family Support

- Responsible for implementing and recommending the Family Support programs
- Responsible for assisting with the organization of the biennial Family Conference
- Responsible for the administration of the Cystinosis Research Network internet support group
- Responsible for welcoming new families by providing means of support such as access to internet support group, CRN website, written materials, referral to other families, etc.
- Responsible for oversight of membership/donor database in conjunction with Development

Education and Awareness

- Responsible for recommending and implementing education/awareness programs for the organization
- Responsible for assisting with organization of the biennial Family Conference
- Responsible for Strategic Alliances and partnerships aimed at enhancing the Cystinosis Research Network’s mission (e.g., NORD, Global Genes, charitable group affiliation, etc.)
- Responsible for Public Relations: publicity, media relations, and corporate communications
- Responsible for publishing the CRN newsletters
- Responsible upkeep and oversight of the organization website

These responsibilities are executed in pursuit of our Vision and Mission:

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

Mission - The Cystinosis Research Network is a volunteer, non-profit organization dedicated to supporting and advocating research, providing family assistance and educating the public and medical communities about cystinosis

The raising of funds is critical for CRN to continue to be able have a positive impact on individuals, organizations and legislation. To achieve this, The Cystinosis Research Network has a Development Committee committed to assisting individuals and families in sponsoring fundraising events. I cannot over emphasize how important is for our community to be self-sufficient and self-advocate. Raising funds to support our programs and activities is one way of advocating for our community. The following are representative of fundraising events that have taken on behalf of CRN since our last newsletter:

livgracefully – held in memory of Livia Grace Stilke and sponsored by Dave Kirsten and Mason Stilke

Long Island Charities Foundation – awarded in honor of Brandon & Johnny Maccarone and sponsored by Peter Ferrandiino, close family friend of John and Arlene Maccarone

Microsoft Give Campaign – held in honor of Evan & Alexis LeBeau and sponsored by John Paolello, Brittney LeBeau’s brother-in-law

May!Hem 2016 – held in honor of Sierra Ayers and sponsored by Julie Ayers

Christmas Palooza For Cystinosis – held in honor of Chandler Moore and sponsored by Clint and Annie Moore
Please reach out and thank sponsoring individuals and families for their commitment and contributions, enabling us to continue to be powerful advocates for our children. Remember, how can we expect others to advocate for us if we do not take a strong stance ourselves?

Lastly, as the year comes to an end, please do not forget to consider CRN for a year-end contribution, AND tax deduction!!!!

I hope each of you give serious consideration to engaging in some capacity with the organization. The Development Committee is available to assist you when you are ready to sponsor an event or join one of our committees. Please reach out to one of our committee members or myself jose.morales01@icloud.com / 203 722-9292, to become involved.

“You can give without loving, but you cannot love without giving.”
— Amy Carmichael

Development Committee
Chair – José Morales – jose.morales01@icloud.com

Corporate Sponsorship
Jeff Larimore - Jlarimore@arnoldfamilycorp.com
John Maccarone - johnm2maccaroneplumbing.com
Deb Reed - dk_reed@hotmail.com

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Katie Morrison - katielmorrisson91@gmail.com
Briana Smythe - bri_ana15@hotmail.com
Jen Wyman - jwyman@comcst.net

Administration
Christy Greeley - Greeleycd@aol.com
Jen Wyman - jwyman@comcst.net

Alex Morales, with her Aunt and Grandmother
At the end October, the Cystinosis Research Network sponsored a gathering for adults with Cystinosis in Charlotte, North Carolina. This was a unique opportunity for sharing concerns and brainstorming topics of interest specific to those of us who have transitioned to adulthood. Eleven adults (including one spouse) with differing backgrounds and childhood experiences met without medical professionals or parents present. In a small period of time, participants were able open up about uncomfortable topics and many topics that haven’t been discussed in a group setting before this session. This forum allowed people to feel safe in speaking their minds, expressing honest and often emotional opinions within a very supportive and uncensored group.

Several projects are beginning based on discussions from this meeting. As the projects are more clearly defined and move forward, we will share them, hopefully very soon!

This is just the beginning. It is like planning for a new building. The first summit developed ideas and initial plans but others are needed to build a firm foundation and get the project underway. Our group was very energized to address issues that will help us and those who follow after us to have more opportunities and a better future.

“I am looking forward to participating with the CRN board for future small gathering events like this one. Intimate events, such as this one are just the beginning of the chapter for adults living with Cystinosis. This is a chapter of strength and optimism.” -Jenn Loglisci

The Future by Design group would like more people to be able to attend and contribute as we move forward. We are hoping to have more summits around the country. So if you are an adult, please join us at future summits to continue our fellowship, education, positivity, and support for each other as we design our future and hopefully improve things for the next generations. If you are interested, please contact Aimee Adelmann at adelmann.aimee@gmail.com.
I had the honor of representing The Cystinosis Research Network (CRN) at two Cystinosis Patient & Caregiver Town Halls: Austin, Texas, June 3 – 4, 2016 and Portland, Oregon, June 10 - 11, 2016. CRN is responsible for presenting an overview of the important role patient advocacy organizations are assuming in the disciplines of research and healthcare. The Town Halls are sponsored by Raptor Pharmaceuticals and focus on providing individuals the opportunity to engage in a content and experience rich two-day program. The program has a focus on the roles of Caregivers/Care Partners, Adults and Teens & Pre-Teens.

**Town Hall Objectives:**

- Improve the support and education resources available to the Cystinosis community
- Identify educational information and tools to support patients and caregivers
- Exchange ideas and information that will help empower people in the Cystinosis community

Key Elements of the Town Hall Programs:

- Cystinosis Resources & Services
- All About Cystinosis
- Managing Cystinosis: Nutrition
- My Cystinosis Journey
- Scavenger Hunt

**- Cystinosis Advocacy**
-Insurance 101
-Thriving With Cystinosis

The CRN's presentation emphasized the importance for all individuals involved with Cystinosis to be active advocates for the community. I utilized CRN as a framework to illustrate how individuals can become powerful advocates in three principle areas: Research, Family Support and Education and Awareness. Both Town Halls were well attended. Our community is indebted to Raptor for creating these great learning opportunities.

Town Halls held in Pittsburgh, Indianapolis, St. Louis, and Boston

By Clinton Moore

It has been a great pleasure representing The Cystinosis Research Network at the Pittsburgh, Indianapolis, St. Louis, and Boston Town Hall meetings. The best memory for me is watching the families interact with each other. Many times they have met for the first time, but seem like they have known each other for years. The patients and caregivers alike seem to learn more from each other than they do from talking to their doctors or researching online articles. I've often listened to families share personal experiences thinking they were the only ones that had experienced that particular situation, but in fact were talking to someone who had experienced the same things.

It was an absolute pleasure attending these meetings and I look forward to continuing representing this great community.
Chandler’s Chance and Nemours Cystinosis Family Educational Day Held on December 3rd

By Clinton Moore

This year we will be hosting our 4th annual Chandler's Chance Fundraiser and the 2nd Annual Nemours Cystinosis Family Educational Day both on December 3rd. It will be a great opportunity for patients and caregivers alike.
Join us for the 2nd Annual Nemours Family Cystinosis Day

We welcome patients and families to join us for a fun filled and educational conference aimed at providing you with the latest updates in the care of nephropathic cystinosis. We are very excited to have a work-renowned cystinosis expert, Dr. Paul Grimm, from Lucille Packard Children’s Hospital as our guest speaker.

Following the event you are welcome to stay for Chandler’s Chance—a Christmas Palooza for Cystinosis. This annual evening event is a fundraiser for the Cystinosis Research Network. It’s a fun filled family event consisting of food, fire truck rides, hay rides, BB gun shooting, Santa Claus and much more!

Date: December 3rd, 2016

Location: Milton CHEER Center, 24855 Broadkill Road, Milton, Delaware 19968

Housing: We have applied for a grant to offer attending families 2 complimentary local hotel nights

Preliminary Agenda:

11:00-11:15am          Welcoming Remarks: Dr Zaritsky
11:15- 12:00pm          Advances in the Treatment of Nephropathic Cystinosis: Dr Grimm
12:00-1:00pm            Lunch/Breakout Session: all
1:00-1:30pm              Eating well with Cystinosis: Megan O’Neill
1:30-2:00pm              Update on Pediatric Kidney Transplantation: Dr LaRosa
2:00-2:30pm              Break/Family Networking
2:30-3:00pm              The Value of Patient Engagement in the Setting of Ultra-Rare Diseases: Dr Zaritsky
3:00-3:15pm              Concluding Remarks: Drs Grimm and Zaritsky

To RSVP please contact Clint Moore at Clintonmoore1@aol.com or call our office at 302-651-4426

This program is supported through an educational grant from Raptor Pharmaceuticals, Inc.
Cystinosis in Emerging Adulthood Article:
Q & A with Dr. Maya Doyle

By Christy Greeley

In 2011 and 2012, the CRN funded a qualitative study based on focus groups and interviews with adults with Cystinosis and their parents. In 2013, the Society for Social Work and Research awarded the study their Outstanding Dissertation award in 2013. To read the full article, visit the CRN website at: https://cystinosis.org/research/article-library/transition

We were able to ask Dr. Maya Doyle some questions about her study:

Q: What was the most surprising finding during your research?
A: There were many surprises during the study, which was originally focused on the transition from pediatric to adult-oriented care. Many of the people with Cystinosis that I interviewed were part of the cohort who had access to cysteamine in a variety of forms before Cystagon received FDA approval, and interviews were done in the years leading up to the FDA's approval of Procysbi. It was a very exciting and anxious time, but patients (and families) were very reflective about what they had experienced growing up with Cystinosis, the strategies families used to manage the disease and find a fit for the illness within the lives, and their sense of themselves. The study also highlighted the ongoing concerns that young adult patients and their families have about the transition to adult-oriented care, and to becoming a more independent adult.
Q: What do you hope families take away from the study?
A: I hope families are reminded of the many strengths that they develop in living with a rare disease, and in being part of this rare disease community. What they shared with me is helping not only people with Cystinosis, but patients and families with other disorders - I have given talks and webinars based on what I learned from all of you, because so many of the challenges are shared. My goal was to bring the voice of patients and families to clinicians and researchers - it’s definitely been heard - but we are far from done. There is always more research and more advocacy to do!

Q: How did you initially learn about Cystinosis? What drew you to working more intimately with the Cystinosis community?
A: The first kidney transplant I ever took care of - in 1999 - was a 13 year old girl with Cystinosis. Transition of care wasn't easy. But she's 30 now and she's fabulous! I got to know the community through her and her mother. I think I was also drawn to this work because of my own experience with type 1 diabetes - which changed from being a fatal disease to a chronic one, which then has presented new challenges as treatments have advanced. I've been part of CRN family meetings since 2005, have been a professional advisor to the Network.

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

Visit http://www.cystinosis.org/families-in-your-area. Send your contact info to the e-mail listed to learn who is in your area. It’s that easy!
Remembering Phil Morrison

Editors note: Former CRN Board member, Katie Morrison, lost her dad last April. The notice was too late to be included in our Spring Summer edition. His obituary is below. To the Morrison Family CRN sends love and condolences.

On Sunday April 17th, Phil Morrison stopped “LIVIN’ THE DREAM”. Phil’s 57 years were filled with many joys and accomplishments. He had a long career in the wireless industry that went back as far as bag phones, car kits and Roaming Guides. During the past several years Phil worked as a professional driver for TN Limo Service. This was a perfect job for Phil. He loved to drive but even more he loved to meet new people and discuss the important things in life like Memphis State Basketball (if you referred to the University as University of Memphis you immediately lost credibility), SEC Football, local politics, growing tomatoes and his children. He was blessed with the Gift of Gab and shared it with many of us. His favorite movie was “Animal House” and he would try to sneak in a line from the movie during his conversations just to see if anyone would catch it.

Phil’s hobbies included reading, listening to music and gardening. He would read several novels a week and was particularly fond of fictional murder mysteries and military espionage novels. When asked about his preference in music he would respond “I like the real stuff, not the crap my kids are listening to.” What he meant was, he liked The Eagles, Little River Band, The Doobie Brothers, Kenny Loggins, Hall and Oates, Jimmy Buffett and Earth Wind and Fire. There is no doubt that he has already talked with Glenn Frey and Maurice White and is convincing them to form the best house band ever. Phil’s gardening skills were beyond words. He once kept track of the cost it took to produce each tomato he harvested. Those were some of the best $16 (each) tomatoes ever grown.

Phil is survived by his two ex-wives (no, this was not the cause of death) Lynette Bobbitt Best of Pace, FL and Melissa Morrison of Bartlett TN. His marriage to Lynette brought two beautiful girls into the world, Alice Kristine Smith (30) (David) of Pensacola FL and Katherine L. Morrison (25) (Marcus Mitchell) of Memphis TN. His marriage to Melissa gifted him with another beautiful daughter Olivia Nicole Morrison (13) of Bartlett TN and a son Jackson Kenneth Morrison (11) of Bartlett. His Father and Mother, Retired Navy Commander V. Eugene Morrison and Jacqueline Morrison of Butler MO. Three brothers Michael Morrison (Cindi Walker) of Houston TX, Mitch Morrison (Patty) of Brentwood TN and Ken Burnette (Susie) of Bartlett, TN. One sister Amy Morrison King (Steve) of Cayuga, IN. Phil loved and was very proud of his nieces and nephews, Alyssa Walker Morrison of Houston TX, Kelsey Burnette of Bartlett TN, John Morrison of Greenville, SC and Clay Morrison of Brentwood TN.
In lieu of flowers, the family is asking that donations be made to either Cystinosis Research Network, 302 Whytegate Court, Lake Forest, IL 60045 or Bartlett Hills Baptist Church Youth Program, 4647 Ellendale Rd, Bartlett, TN 38135.

Meet new friends
Reconnect with old ones
Share your journey with those who “get” it
2017 CRN Family Conference
July 13-15
Snowbird, Utah

See you there!!
Join the Cystinosis Research Network

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

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

“Dedicated to a Cure. Committed to our Community”...whether you are...

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

Joining the Cystinosis Research Network enables you to:

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

Join Cystinosis Research Network today!

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

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

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International: (Including Canada) Base rate (see above categories) plus $10.00 for postage.

Payable in US dollars

Please complete the form & mail with check payable to CRN to:

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

Name________________________________________________________________________
Street________________________________________________________________________
City & State_______________________________________ Zip Code__________ Country ____
Phone__________________________Fax_______________________Email________________

Name of Child / Adult / Acquaintance / Patient affected with cystinosis:________________________

Join A CRN Support Group

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

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

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

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

To join a support group, visit www.cystinosis.org.
YES, I want to help children and adults with cystinosis.

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

Name__________________________________________________________
Street_________________________________________________________________
City & State_______________________________________ Zip Code_________________
Phone____________________Fax____________________Email____________________
In Honor Of_____________________________________________________________
In Memory Of___________________________________________________________
You may send notification of my gift to:
______________________________________________________________________

Please check all that apply:

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

Search the Web with GoodSearch & Raise Money for CRN

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

To get started, go to goodsearch.com and select Cystinosis Research Network where it says "Choose your cause." Then search like you normally would!

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

Make Purchases at GoodShop & Raise Money for CRN

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

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

GoodShop will donate up to 30% of your purchase to CRN. Some of the hundreds of retailers include: Best Buy, iTunes, Home Depot, Amazon, Barnes & Noble, Dell, Banana Republic, Macy's, Target, Wal-Mart, Ann Taylor Loft, Chicos, Coldwater Creek, American Eagle Outfitters, and many more!
# United Way Contribution Guidelines

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

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<tr>
<td>Address</td>
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<tr>
<td>Telephone</td>
<td>1-866-276-3669 (toll free), 1-847-735-0471</td>
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<tr>
<td>Web Page</td>
<td><a href="http://www.cystinosis.org">www.cystinosis.org</a></td>
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The local United Way organization will contact The Cystinosis Research Network via phone, fax, or e-mail to request we prepare and submit documentation verifying our status as a non-profit organization.

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

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

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

## Donate to CRN by Selling on eBay

CRN is registered with MissionFish, the exclusive charity provider for eBay Giving Works. eBay sellers can now list items through eBay Giving Works and designate a percentage of the sales to go to CRN. The seller picks the percentage, and all money donated is tax deductible. eBay will even refund a percentage of listing and final value fees that is equal to the percentage sellers donate! Items listed with eBay Giving Works are given a special icon, so they stand out. Some sellers report 20-40% higher sale prices for the exact same item using eBay Giving Works. Give it a try, and be sure to tell established eBay sellers about this great opportunity to give to CRN!

Learn more about the CRN Amazon Smile Program by clicking on the link below:

Cystinosis is a rare, genetic, metabolic disease that causes an amino acid, cysteine, to accumulate in various organs of the body, including the kidneys, eyes, liver, pancreas, brain and white blood cells. Without specific treatment, children with cystinosis develop end stage kidney failure at approximately age nine. The availability of cysteamine medical therapy has dramatically improved the natural history of cystinosis so that well treated cystinosis patients can live into adulthood.

CRN’s Vision and Mission

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

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