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2017 CRN Family Conference "Soaring to New Heights" a Great Success!



Children and adults living with Cystinosis attend the 2017 CRN Family Conference held in Snowbird, Utah in July.

The 2017 Cystinosis Research Network (CRN) Family Conference was held July 13 - 15, 2017 at the Snowbird Ski and Summer Resort in Snowbird, Utah. It was a spectacular venue where approximately 280 family members, physicians, researchers, and volunteers attended from 32 states and 6 different countries, including 65 families and 55 individuals with cystinosis. All attendees were able to learn about the latest updates in cystinosis research through lectures, workshops, poster sessions, and medical and

patient panels. There were many opportunities to socialize with other patients and families, which provided valuable connections and friendships that will last a lifetime. The conference theme, "Soaring to New Heights" was clearly appropriate as the concentration of the event was on demonstrating ways in which the community continues to move onward and upward. "Camp CRN" was a new addition this year featuring programming specifically designed for children, tweens with cystinosis, teens with cystinosis, sibling

Continued on page 8



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The President's Letter

By Clinton Moore

Sitting in a semi-quiet hospital room, after yet another one of my son's numerous surgeries, doesn't seem like the perfect place to be writing this. I mean with the constant ins and outs of the nurses, doctors stopping by, and the never-ending beeping from the IV pump, who could possibly concentrate? Luckily this time it's a simple surgery, with just tonsils being removed....but with cystinosis....is anything really "simple"?

Every time we return to our "home away from home" I'm taken back to the earlier years when we were trying to get diagnosed. After many improvements and a new inpatient wing added to the hospital, the room in which we first heard the word "Cystinosis" is no longer a patient room. Today it's just used for storage and possibly office space down the road, but I can still see it just the way it was twelve years ago. I believe I could tell you word for word every question and conversation that took place in that room during that week and a half. We didn't know it walking into that room that day, but absolutely every part of our life was about to take a dramatic change once we walked out.

Since then we have been back to this hospital what seems like hundreds of times. So many appointments with all of the "ologists", some routine check-ups, some not. This is where we have met many people, shared many laughs, and even more tears. This is where we have searched for answers that we thought we would never find. This is where we have received news that seems to stop your heartbeat for a moment or two. This is where we have been forced to hold hands as a family and make decisions that we could only hope were the best. As a grown man I don't mind telling you...this is the place that has had me on my feet running...and the same place that has knocked me to my knees to pray. So, perhaps, this is the perfect place to be writing this after all.

The Cystinosis Research Network is totally committed in assisting families that are going through any part of this journey. In this edition of the Cystinosis Advocate, you will read about the many involvements of the Cystinosis Research Network, from funding research, to education and awareness, to family support and much more.

As your newly appointed President, I am committed to continuing the hard work and progress already established by those before me. The past few months have been challenging as I have been getting acclimated and learning this new role. I suspect the next few years will be just as challenging as cystinosis and all the aspects of it can be



quite complex. Now is the time to continue the forward momentum, to strengthen relationships both nationally and internationally, create needed programs, improve existing programs, and to continue to educate and advocate at every given opportunity.

The Cystinosis Research Network is YOUR advocacy group. Use it to your advantage. If you ever have any questions, concerns, comments, or if the CRN can assist you in any way, please reach out to myself or any board member as we are very willing to help or assist in getting you whatever it is you need.

Sincerely,
Clinton Moore

Clinton Moore Offers a Unique Pledge to the Cystinosis Community

The cystinosis community is often viewed as a very tight knit community, with all the families sharing experiences, worries, information, and prayers with each other on a near daily basis. I have often received such support myself from many members of this community in my time of need. I am often awestruck when I see just what extent people will go to, to lend a helping hand, and personally I would want it no other way. What better way to travel this journey than with so many supportive people that know exactly what you're going through and are willing to hold you up when needed?

With all the possible complications and issues that cystinosis can bring, no one ever wants to hear the word "cancer" among everything else they are dealing with. No one wants to hear that word at all, period. Unfortunately, this has been just the case for some members of this community. Cancer brings on a whole new batch of worries, fears, medications, and unknowns. The medication alone can be terrible to deal with and has many adverse side effects. This "new" diagnosis can lead a person to feel very alone, but we here at the Cystinosis Research Network want to change that.

You are never alone in any part of your cystinosis journey, or any road you are forced to travel down. To show our support to you, We decided to shave my hair totally off, and will continue to do so every October from now until cystinosis no longer affects a single family. If you have received a cancer diagnosis or any issue with cystinosis at all, please reach out to the Cystinosis Research Network. We are here to help. We are your advocacy group.

Best Wishes,
Clinton Moore



Clinton Moore shaves his head in solidarity and support of those in the cystinosis community battling cancer.

CRN Announces Two 2017 Academic Scholarship Recipients

By Terri Schleuder

The Cystinosis Research network is pleased to announce our 2017 Academic scholarship recipients. We had several very qualified applicants this year making our choice a difficult one. The winner of the 2017 CRN Academic Scholarship for an Individual with Cystinosis for \$1,000 is Emily Patterson. Emily will begin her college career at Central Carolina Community College focusing on early childhood education.



Congratulations to Emily Patterson, the recipient of the 2017 CRN Academic Scholarship for an Individual with Cystinosis for \$1,000.

Emily states, *"It is an absolute honor to receive the 2017 CRN Academic Scholarship for an Individual with Cystinosis. This scholarship will help me obtain my certificate in Early Childhood Education and, ultimately, fulfill my goal of working with children in an educational setting. As most of you know, 2017 has been quite a busy year for my family and me. In February, I was blessed to receive a kidney transplant. However, due to multiple complications, my hospitalization was extended. That, along with the crazy hectic post-transplant schedule, made it hard for me to even begin thinking and planning for college and financial aid. I can't express my gratitude enough in this short paragraph. All I can do is say "thank you." I don't know what the future holds but I am looking forward to seeing where it takes me. Your generous contribution is much appreciated and will be put to good use."*

Tyler Morrill was the recipient of the 2017 CRN Academic Scholarship for the Sibling of an Individual with Cystinosis. Tyler will begin his college career at Miami of Ohio with a long term goal of a career in the healthcare field.



Congratulations to Tyler Morrill the winner of the 2017 CRN Academic Scholarship for the Sibling of an Individual with Cystinosis for \$1,000. Tyler is pictured here with his sister Megan Morrill.

From Tyler, "My name is Tyler Morrill and I am the proud recipient of the Sierra Woodward Sibling Scholarship Award. I am just beginning my freshman year at Miami of Ohio. I am starting out in the Kinesiology department and have plans to go into Physical Therapy. Your award will go toward my tuition. I have grown up knowing how important the Cystinosis Research Network is to my sister and feel very happy knowing this amazing organization reaches out to the extended family. Thank you again for helping me achieve my goals."

Both of these fine young people demonstrated excellence in academics, extracurricular activities and leadership skills and were highly recommended by those writing letters on their behalf.

We wish them the very best as they continue to work on the collegiate and life goals.

News for CRN Families and Genetic Testing

DNACheckup (dnacheckup.org) has launched its carrier genetic testing for families affected by recessive genetic disorders.

DNACheckup is the first and only genetic testing service that:

- 1) is affordable (\$200 all inclusive);
- 2) is online physician-ordered; and
- 3) allows participants to order the tests and collect the saliva samples from the privacy of their own homes. We are testing for only mutations (usually 1 or 2) already identified in the affected family member, and hence the test is personalized for each family.

Learn more at the link above.



CRN Welcomes New Board Members and Current Board Members to New Roles

CRN is pleased to announce former board member, Jen Wyman is returning to the Cystinosis Research Network executive committee as the VP of Family Support replacing Clinton Moore in that role. Clinton assumed the position of CRN's President at the end of Jeff Larimore's term in August. In addition, current board member Ina Gardener has assumed the Secretary's position.



Jen Wyman graduated from Albion College with a Liberal Arts Degree, majoring in English and Elementary Education. While she was teaching in Tecumseh, MI she received her Master's Degree in Educational Psychology. She and Tim married in 1992, started a family in 1994 and moved to Bloomfield Hills, MI. It was then that she became a stay at home mom. Kacy was diagnosed in 2006. Almost immediately after that, their family became involved with CRN and Jen signed on as a board member. She has served as the VP of Development in the past and is looking forward to serving as the VP of Family Support. Tim and Jen have been married for 25 years, still live in Bloomfield Hills, MI and have three children, Matt (23), Jack (21) and Kacy (15-with cystinosis).

We are also welcoming three new CRN board members, **Aimee Adelmann, Jill Emerson, and Larry Thomas**. Please read below to learn more about these dynamic individuals.

Aimee Adelmann has over 15 years of experience in non-profit healthcare and support industries and has spent last four years specifically targeted toward education related to donation and transplant. Her experiences in healthcare isn't only as a professional, but also as a cystinosis patient and two-time kidney recipient. As Director of Education and Outreach at Donate Life Northwest she has been able to combine her professional and personal experience with a passion to help educate people throughout Oregon, SW Washington, and nationally on the great importance of organ, eye, and tissue donation.



Aimee is excited to use her experience to help bring together our adult population and advocate for cystinosis patients and their families. With the new initiative Future by Design, she hopes that adults with cystinosis can come together and bring our voice forward to educate, advocate, mentor, within our community and throughout the public.

In her free time Aimee enjoys volunteering with various organization across the nation including, Cystinosis Research Network and Donate Life America. She also is a public speaker on various health related topics, enjoys baking and cooking, loves to travel, and like a true Oregonian enjoys the outdoors.

Aimee Adelman email: adelman.aimee@gmail.com

Jill Emerson graduated from the Pennsylvania State University with a Bachelor of Science degree in Accounting and Saint Joseph’s University with a Master of Science degree in Business - Human Capital Management.

Jill has spent over twenty years working in the accounting industry and is a Certified Public Accountant in the state of Pennsylvania. Currently, she is a Manager at Your Part-Time Controller LLC, an accounting firm that works with nonprofit organizations in the Philadelphia, Washington DC, NYC and Houston areas. In this role, Jill works closely with nonprofit organizations, their Boards of Directors and auditors, and provides them with high-level accounting and finance services. In addition, she is a member of the organization’s internal Quality Control Review team. Jill has also served as the Director of YPTC’s Bookkeeper Technician University, a program that provides pro-bono bookkeeper training to nonprofit organizations.



Jill resides in Hammonton, New Jersey with her husband, Clay, and their three-year-old daughter, Brooke, who was diagnosed with cystinosis at 16 months of age. In addition to being “Mommy”, Jill enjoys soccer, yoga, reading, cooking and camping, kayaking and fishing with Clay and Brooke.



Larry Thomas lives in Orchard Park, a suburb of Buffalo, New York. His son Garrett, is a 21 year old Junior in college and has cystinosis. Larry is a Recruiter in the Healthcare IT space and is fortunate enough to work from home at a job he loves. This is Larry’s first term on the board and he is looking forward to whatever he can do to help CRN.

Congratulations to all, we are pleased and excited for the new ideas and energy this group brings to CRN.

Family Conference Continued from Page 1

tweens and sibling teens.

A new twist to this year's conference was the availability of live streaming for some portions of the conference. If you missed the live stream event, you can still view recordings of most of the presentations on the CRN YouTube channel at https://www.youtube.com/channel/UC3BCCVrDmY6M7ZKfUeBmOYQ/videos?disable_polymer=1

With the support of our generous sponsors, Horizon Pharma, Leadiant Biosciences, and Eloxx Pharmaceuticals, CRN was able to provide three days of programming which was aimed at both families and professionals.

Wednesday began with the CRN Board of Directors meeting. On Thursday, conference registration began. The CRN Scientific Review Board also met on Thursday to review this year's Call for Research Proposals submissions. An announcement regarding studies funded for 2017-2018 can be found in the Research Update section of this newsletter. Families



Dr. Elena Levchenko, Dr. Neveen Soliman, and Dr. Ewa Elenberg

were welcomed to the conference on Thursday evening with a reception which was highlighted by the kickoff of this year's Snow City Arts project in which individuals created representations of themselves as a bird in mixed media sculpture forms. Each bird was uniquely designed and were suspended together to create a collaborative time based hanging art installation of birds flying in an upward spiral. The upward spiral form was chosen to signify how those impacted by cystinosis can reach new heights working as one community. Finally, families had an opportunity to share their journeys with one another on Thursday evening during the Family Introductions Session.

Friday was a full day focused on increasing our understanding of cystinosis and relevant medical discussions concentrating on infant, transition and adult issues. The day kicked off with an outstanding keynote address from Jerry Cahill. Jerry is a renowned advocate in the cystic fibrosis community. As a CF Ambassador at the Boomer Esiason



Snow City Arts project, mixed media bird sculpture, created by attendees.

Foundation, (BEF) Cahill leads its scholarship and grants division and is the founder of Team Boomer, the athletic arm of BEF dedicated to encouraging exercise among CF patients while simultaneously raising money for scholarships. He has also started the program "You Cannot Fail". His message of hope and perseverance for those living with chronic illness was inspiring. The rest of the day included a review of cystinosis old and new by Dr. Zaritsky and a talk on the state of the world of Cystinosis by Dr. Langman. John Ray gave an extensive review of how to be an effective advocate in Washington, D.C. Panels were conducted in the afternoon which were moderated by the CRN Advisory Board members and other experts in attendance on issues related to the management of infants and children, adolescents and adults, social relationships and gaining independence, self-management and medication adherence for adults, Education/IEP issues, transplant and dialysis, and depression and mental health for adults. Finally, the medical panel highlighted all the medical professionals in attendance, and

audience questions were addressed. To wrap up the day, a Meet and Greet family dinner was hosted by the CRN Future By Design group of adults with cystinosis whose goals include providing support and encouragement to young adults, parents and children living with cystinosis.



Dr. Stephanie Cherqui, presents her exciting research on stem cell gene therapy for cystinosis.



Dr. Jennifer Simpson describes the nanowafer, a novel and innovative therapy for corneal cystinosis.

Saturday began with updates about Cystaran by Leadiant Biosciences and Walgreens Specialty Pharmacy and Procysbi by Horizon Pharma. A Research Update session was added this year where cystinosis researchers gave brief summaries of their current work. These included talks by Dr. Cherqui on Stem Cell Gene Therapy, Dr. Simpson on the Cysteamine Nanowafer, Dr. Levchenko on the Zebrafish model and successful fertility treatment in males with cystinosis, Dr. Langman on the history of Procysbi studies, Dr. Elenberg on chronic kidney disease, dialysis and transplant outcomes in patients with cystinosis, and finally Dr. Nesterova on bone deformities in patients with cystinosis. Recordings of most of these talks can be found on the CRN YouTube channel referenced above. The afternoon featured a poster session which showcased a mix of science, medicine, industry and advocacy group and patient experiences. Twelve posters were featured, including many highlighting CRN funded research. To wrap up the day we enjoyed interacting during the "Parents of Children and Adults with Cystinosis Panel", where

parents of children and adults with cystinosis answered questions and shared their experiences, and the "Future by Design" presentation where teens and adults living with cystinosis shared how they live their lives to their full potential given the challenges they face. There was also a closed panel moderated by Dr. Levchenko on reproductive issues for cystinosis adults and their partners. Finally, everyone said goodbye during the Farewell Dinner Dance, celebrating the community soaring to new heights!

An announcement will be made very soon on the location for the next CRN Family Conference, to be held in July 2019 – stay tuned for more information! We look forward to gathering together again soon!



Dr. Doris Trauner, Christy Greeley, Dr. Bill Gahl, Dr. Jennifer Simpson, Dr. Ewa Elenberg, and Dr. Katharina Hohenfellner.

2017 CRN Family Conference The Highlights!



Dr. Joshua Zaritsky, Chief of Nephrology at the Nemours/Alfred I. duPont Hospital for Children.



Nancy Parsons, Vice President of Pharmaceutical Products at Leadiant Biosciences, Inc.



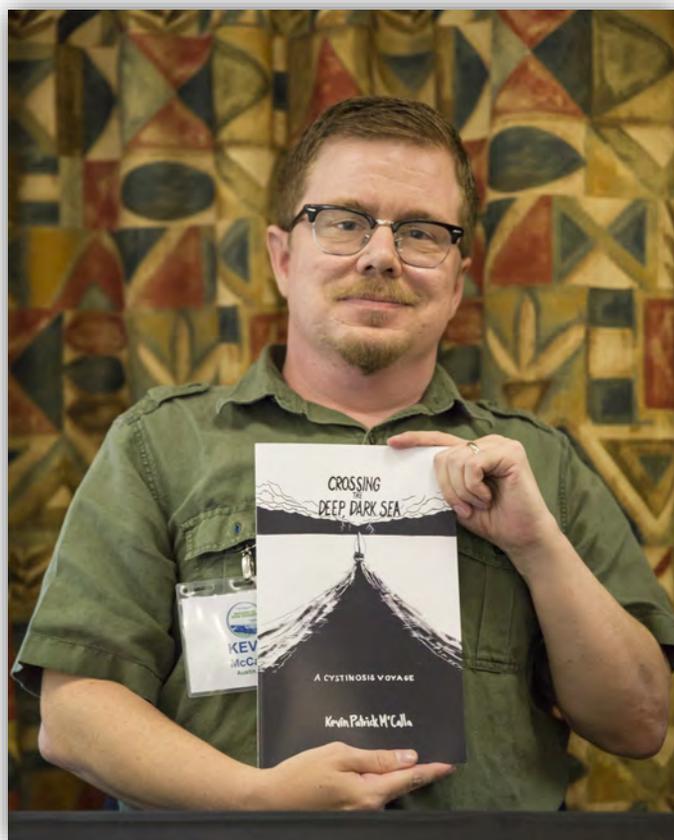
Robert Metz, Senior Vice President, Business Operations and External Affairs, Horizon Pharma.





Above: Garrett Thomas, Sara Healy, Emily Mello, and Jana Healy our Cystinosis Superheroes bring fun and excitement to Thursday's Welcome Reception and the Family Introductions.

Right: Graphic artist, Kevin McCalla debuts his 2nd Cystinosis book, a compelling reflection of living life with Cystinosis. He signed many copies for attendees.



CRN President elect, Clinton Moore



CRN executive director, Christy Greeley and Lesli King with Leadiant Biosciences



front: Marcus Root. back: Zander Vachon



Glass artist Lee Knaus, selling his beautiful ornaments with some of the proceeds benefiting CRN.



Claudia Sproedt, Anne Marie O'Dowd, Christy Greeley, Pam Woodward, and Christian Sproedt





Top: Keynote speaker and Cystic Fibrosis advocate Jerry Cahill
Left: Dr. Craig Langman
Bottom: CRN President Jeff Larimore, and Dr. Patrick Gipson



More Highlights



Dr. Bill Gahl



Rebekah Palmer

Far right: Logan Vachon
Right: Dakota Roe



Back row: Shea Hammond, Colleen Hammond, Dr. Elena Levtschenko, Dr. Craig Langman. Front row: Dr. Joshua Zaritsky, Phyllis Kaskel, and Dr. Rick Kaskel

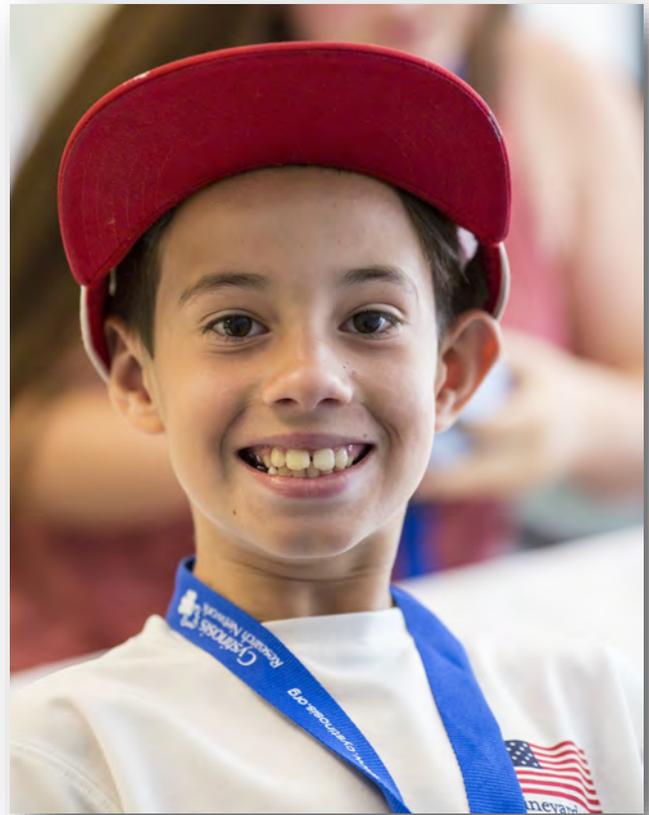


The Sigler family





Terri Schleuder and Clinton Moore at the poster session.



Elijah Mello Teixeira



Ethan Krenzer



Jim, Melea and Claire Martin



Kristina Broadbelt and Tricia Miullins



Larry and Garrett Thomas



2017 CRN Family Conference Attendees



Serena Waldron

Thank You!

CRN offers our deepest and sincerest appreciation to our Corporate Sponsors. Without their generous financial support this conference would not have been possible to plan, organize or execute. We are incredibly grateful for their continued support to our organization. Together we can truly "Soar to New Heights".



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CRN also thanks the families listed below who contributed to the success of this year's conference with their generous donations. We are very appreciative.



Adelmann Family Trust
Birch Family
Hammond Family
Julian Family
McCalla Family
Sexstone Family
Sproedt Family

Cystinosis **Impact Program** Meeting Held in Ypsilanti, Michigan, Sept. 30th

By **Terri Schleuder**

Four families connected by cystinosis, met on a picture-perfect Fall day in the Ann Arbor/Ypsilanti, Michigan area. At the beautiful Eagle Crest Conference Center in Ypsilanti, Michigan, we were able to meet each other, visit, shared stories, and learned more about this disease that has affected all our lives.

The morning provided an opportunity to share how cystinosis had touched our lives, including the positive impacts that having this disease or caring for someone with it has made on each of us.

After a delicious lunch, we listened to Dr. Jeanine Jarnes, pharmD, BCOP, BCPS, from the University of Minnesota. She gave us a clearer understanding of cystinosis as a lysosomal transport disease, and how various therapies work to treat it. We also learned about the many resources available to assist with insurance, medication, finances, doctors and other questions cystinosis patients and families face.

Our family enjoyed meeting some families we had not met before, learning valuable information and connecting in a new way to our cystinosis community.

This four-hour meeting was one of several **Impact Programs** offered across the country in 2017 providing the chance for families to connect as smaller groups in regional areas. Impact meetings in 2017 were held in several locations including Denver, Chicago, Seattle and Annapolis in addition to our area.

More will be offered in 2018.

If you would be interested in attending an **Impact Program** meeting next year, or want earn more about this opportunity, please contact Jennifer Caughlin, at jennifer@caughlinresearch.com (602)793-2552.



Cystinosis families gather in Ypsilanti, Michigan for an Impact Program on Sept. 30th.

Front row: Stephanie Trudell Smith, Jenn Gray, Steve Schleuder, Emma Halsteld, Molly Dunlap

Back row: Carl Schleuder, Terri Schleuder, Jody Simon, Linda Conklin, Jennifer Dunlap

My Journey Living with Cystinosis by Eddie Langley

By Eddie Langley

My name is Eddie Langley and cystinosis has been a part of my life for almost 39 years. I was diagnosed around three months of age only because my parents recognized the signs my older sister had when she was born in 1972. She passed away sometime in 1981.

While I was growing up my parents raised me as "normal", as

they could. The only thing they told me was I had to take my medicine to be able to do the things I wanted. During my younger years I did not have any issues making friends, but had more of an issue with parents thinking because I had a rare disease their kids would catch something.

There were many hurdles I had to jump through throughout life to be successful. Of course there were the multiple doctor appointments, hospital stays etc. Those became routine and I was use to them since I have been going since I was an infant. The harder issues were more on the social side. Bullying entered my life around 5th grade but I was taught to stand up for myself and I did so. This did not last very long. I even made some friends this way and a lot of kids came to me sometimes just to talk and get things off their chest. I still remember when I broke my word to a friend when I told the school about his abuse and never saw him again and all the teacher told me was Charles was now safe.

With the cysteamine in liquid form I always had bad breath and body odor. I had to deal with a lot of heckling, no matter how many times I brushed my teeth, used mouth wash or breath mints. When I was starting to go through puberty I became terrified of girls just because of the odor issues. All my friends understood about the bad breath, but I



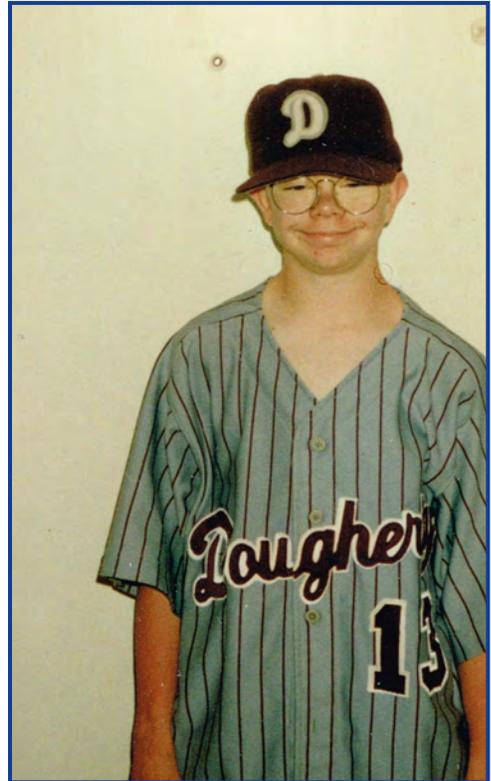
Eddie's first Karate tournament.

still had someone trying to make jokes on the bad days when the cysteamine stench was just extra aggressive. I thought no girl would want to talk to me, so I would literally just run away.

During my childhood and teen years, when people asked why I was so short I would just tell them I have kidney problems and had to take so much medicine that was why I was shorter. That worked great for an explanation. Everyone was surprised since I was so active, they had no idea. This still happens today.

I played baseball from T-ball all the way to making varsity on the high school baseball team. I was involved in martial arts, boy scouts, cross country, chorus, and drama club. In high school I also participated in VICA and took 3rd place in the state for my first culinary arts competition.

My parents and doctors stressed the importance of education and just because I had health issues that was not an excuse to not graduate and try to become an independent adult. I was told that it was necessary so I could continue to receive the quality healthcare I needed for cystinosis. Without corporate medical insurance I would not be able to receive medical benefits due to a pre-existing condition, and



Eddie in his baseball uniform his junior year of high school after a tough game against a school rival.

my quality of life would drastically fall and when my parents passed away if I did not know how to care for myself. I am thankful for my parents raising me to be independent even though I was probably a big pain in the rear during my teenage years. Because of them I have experienced so many things that otherwise I would not have been able to.



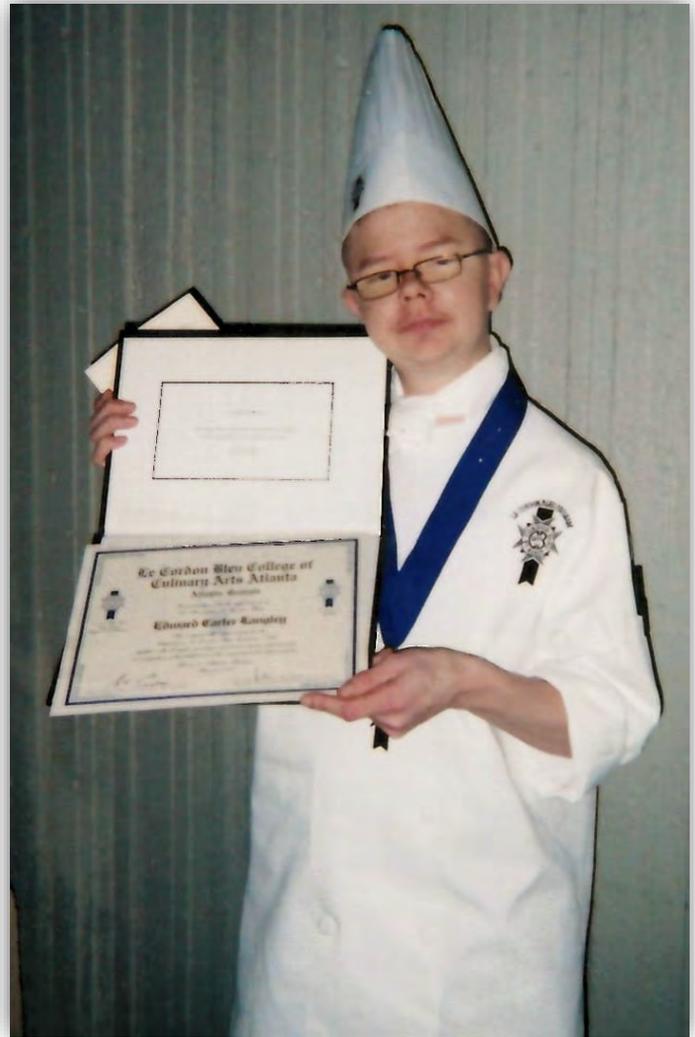
Eddie ran Cross Country one year post transplant in the 8th grade.

Participating in research as a child, and seeing the improvements in treatment throughout the years has helped me along my own cystinosis journey. Having parents who did not let me use cystinosis as an excuse for what I can't do, but instead to focus on what I have to do to be successful was vital. These were common knowledge things like taking my meds on time and not skipping doses, speaking up for myself, listening to the doctors and don't make excuses.

The transition from my teenage age years into adulthood were very eye opening. After graduating high school I started college full-time while keeping a full-time job for insurance, handling the medical bills and the mistakes by both hospital and insurance companies, keeping up with refills for medications, and well just anything that someone with a with a rare disease has to keep up with. Then there are all the adult issues everyone has to deal with. It sure adds up for someone first entering adulthood.

Even though it felt like the weight of the world was on my shoulders, with support from my family and friends, I was able to move forward in my adult life. I started out in casual dining and worked my way to fine dining, and professional ACF competitions where I placed 4th and 3rd out of many competitors. After many interviews and background checks, I was accepted as a contestant on the Hell's Kitchen television show. After the show I continued to work and was recruited by one of the top resorts in the world. I finally got over my fear of women and had my first date when I was 20 years old and got married at 30 years old. The marriage ended in divorce after 2 years.

Within the cystinosis community, I've been very proud to go to the town halls, be a member of Future by Design adult group and travel to Washington to speak to my state representatives on behalf of cystinosis and the rare disease community. I've been able to do all this because my parents taught me not to use cystinosis as an excuse because the world around you doesn't care that you have a rare disease. If you



Eddie graduating from Le Cordon Bleu College of Culinary Arts.

want something, you need to go and get it. Through my years with cystinosis I have been through three kidney transplants, an aortic root aneurysm repair, several peritoneal dialysis catheter placements and removals, an unsuccessful fistula placement, a graft placement, an eye biopsy and countless other minor procedures.

You can continue to move forward in your life even with the bumps in the road that come your way like these. I know all cystinosis people have a different degree of the disease, side effects and difficulties, but I challenge everyone to live life to your full potential. To parents, "try to get your kids involved in activities and start to build their confidence up to help them succeed in their difficult journey that this terrible disease has set for them". Remember cystinosis is a part of your life; nowhere near the whole thing.



Eddie and Dr. Elena Levchenko at the 2017 CRN Family Conference in Snowbird, Utah, July 13-15.

Prayers and good vibes to all,

Eddie Langley

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!

Running for Cystinosis

Learn more at: <https://cystinosis.org/running-for-cystinosis>

What would motivate someone to train for a half marathon in the Texas heat? A cure for Cystinosis!

Amanda grew up watching her little sister, Chelsea, struggle with this rare disease. After diagnosis they moved across the country in search of better care. Chelsea was in and out of hospitals for six years and ultimately needed a new kidney. The impact cystinosis has had on their entire family is immeasurable. But, a kidney transplant and wedding later, Chelsea is 24 years old and doing great! She has inspired Amanda, their mother Holli, and others to join this race on behalf of all those living with cystinosis.



Amanda says, "Having a sibling with cystinosis and knowing other families who are affected by the disease, makes my decision to raise money for CRN that much easier."

Help move the team closer to their fundraising goals and a cure for cystinosis. Tax-deductible contributions can be made online or by check. When sending a check, please be sure to note "SPI Half Marathon" made out to the Cystinosis Research Network, 302 Whytegate Court, Lake Forest, IL 60045.

We were hoping to expand our group of runners for the South Padre Half Marathon on November 12, 2017. For details, visit runspi.com or the "Running for Cystinosis" Facebook page facebook.com/RunningSPIForCystinosis

Goal: \$1,500

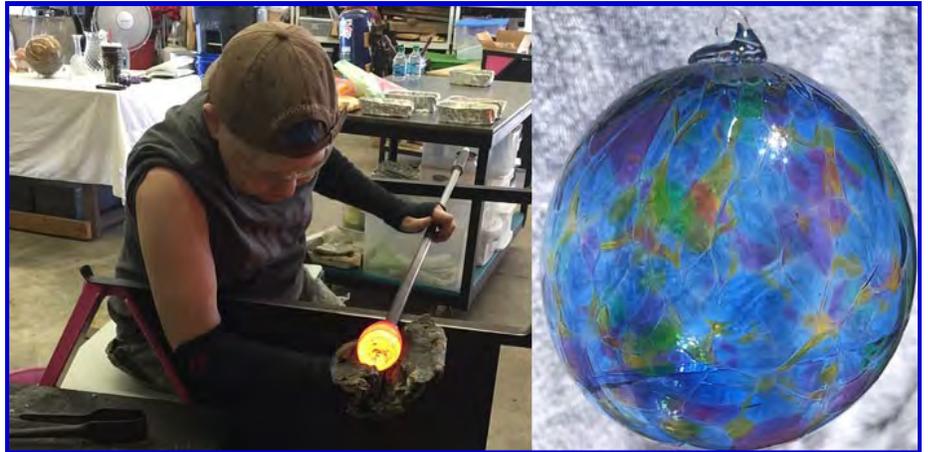
Reach goal: \$2,500



Handcrafted Ornament Sale

Supporting the Croce Memorial Scholarship Program

Help patients in need. Through our ornament fundraiser, you'll support the Croce Memorial Scholarship Program assisting families attend future CRN Conferences. Artist Lee Knaus, a young man living with cystinosis, meticulously molded each item in partnership with Burnt Glassworks Studio in Jacksonville, Florida. The ornaments are ready to adorn your home and the spectacular sun-catching colors and size (5" in diameter) make them the perfect gift!



Glass Artist Lee Knaus is hard at work creating his unique and beautiful glass blown ornaments.

Cost: \$30.00 plus shipping (\$3.00 U.S., \$10.00 outside U.S.)

Orders open through December 1, 2017 based on availability.

The Cystinosis Research Network is a tax-exempt organizations granted "501(c)(3)" non-profit status by the I.R.S., The CRN Federal tax id # is 04-3323789. All Contributions are Tax-Deductible.

Learn more on CRN's website: <https://cystinosis.org/handcrafted-ornament-sale-1>

Breaking News 'Cystinosis Outreach'

A program offering Conversation * Community * Connection

Brought to you by CRN'S **Future by Design**, a group of adults living with cystinosis.

For more information contact Christina20148@hotmail.com

Note: if you are 21+ living with cystinosis and would like to be part of **Future by Design** please contact Amiee Adelman: Adelman.aimee@gmail.com

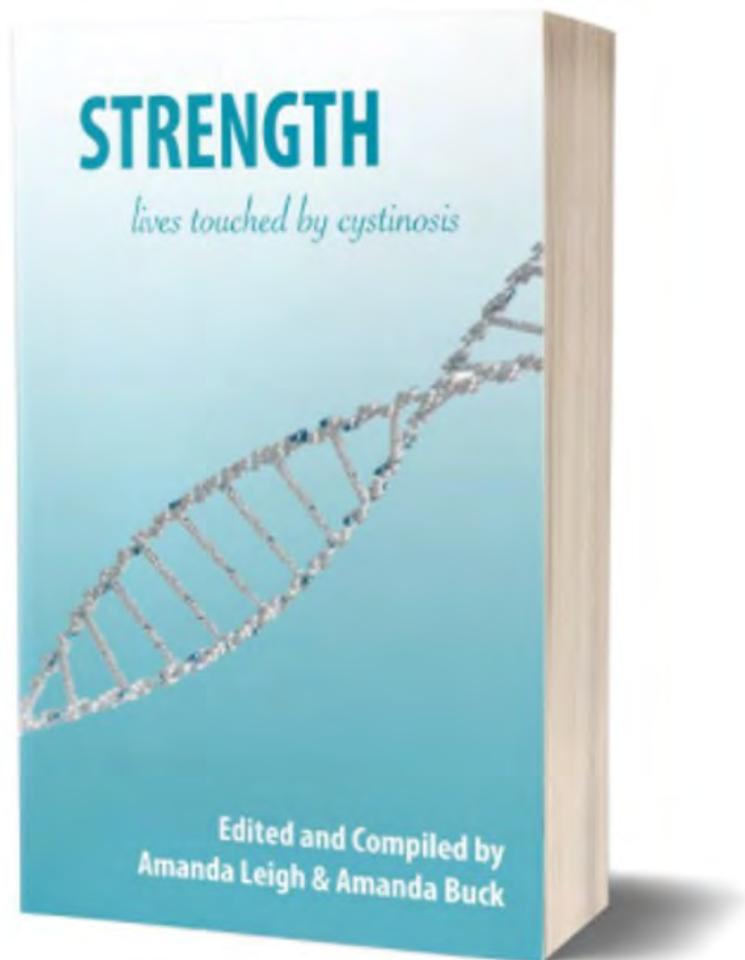
Strength: Lives Touched by Cystinosis

Learn more at: <https://cystinosis.org/fundraiser/book-sale>

Each cystinosis journey is different. However, this collection of stories reminds us of a trait many have in common: Strength.

Hear from over 20 individuals and loved ones impacted by cystinosis. Amanda Buck (cystinosis caregiver) and Amanda Leigh (adult living with cystinosis) deliver this labor of love on behalf of the cystinosis community, but it is intended for EVERYONE who has ever experienced the great pains and joys of life.

Please consider purchasing a copy today. All proceeds benefit the Cystinosis Research Network and move us one step closer to finding a cure. Available through [CreateSpace](#) and [Amazon](#).



ATS Holds First Transplant Patient Summit in Washington, D.C.

By Marybeth Krummenacker

The American Society of Transplantation recently held their first Transplant Patient Summit in Washington D.C. at the historic Mayflower Hotel. Marybeth Krummenacker attended the event as one of over 100 advocates from the United States who were selected through a grant process. Marybeth represented the cystinosis community as the caretaker and advocate for her daughter Laura, who is an 18 year kidney transplant survivor. Attendees included not only kidney transplant survivors, but heart, lung and various other organ transplant survivors as well. As Marybeth said it was amazing to hear how many people have survived for so many years with multiple organs, but the longevity of these organs was astounding. Many were survivors of over 30 years! This is a testament to research and development of new and improved treatments and medications. The hope and the goal for the two day meeting was "One transplant for Life"... and the discussions were very much centered around the process of designing research priorities that include the clinician, scientist and patient, but for the first time to also include the ADVOCATE...and often times the caretaker.

A wonderful part of the agenda was the opportunity to hear from to Rep. Jerrold Nadler of New York who spoke about the bi-partisan support (along with Rep. Raul Ruiz of California and Jaime Herrera Beutler of Washington) for the Living Donor Protection Act. (HR 4616). This important bill prohibits discrimination based on an individual's status as a living organ donor in the offering, issuance, cancellation, coverage, price, or any other condition of a life insurance policy, disability insurance policy, or long-term care insurance policy. The bill amends the Family and Medical Leave Act of 1993 to specifically include living organ donation as a serious health condition that entitles a covered employee to leave under that Act. Everyone was encouraged to contact their local officials to make sure they have signed on to this extremely important piece of legislation. It was an extraordinary morning, especially to hear Congresswoman Herrera-Beutler speak about her infant daughter who was born with no kidneys. The baby survived because of the team of doctors who not only took care of her, but actually worked together to help her thrive and grow until she was big enough to receive a successful kidney transplant from her father. Congresswoman Herrera-Beutler was passionate when she spoke about this important piece of legislation because she has lived it.

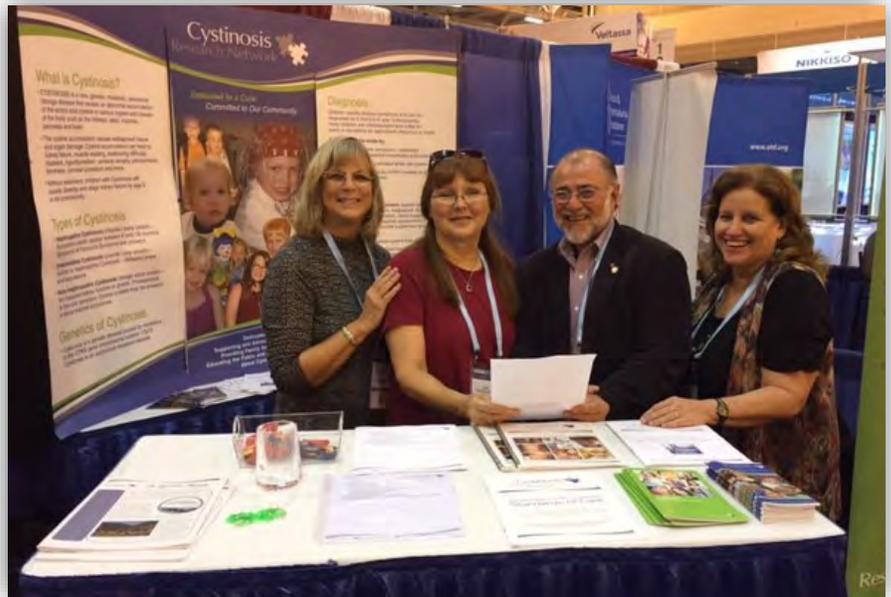
It was a wonderful opportunity to network with advocates and caregivers in the world of organ donation and to listen to and learn from these remarkable and inspiring stories.



CRN Exhibits at the 2017 American Society of Nephrology Conference in New Orleans

By Terri Schleuder

This year was another successful exhibiting experience for CRN at the 2017 American Society of Nephrology conference (ASN) held at the convention center in picturesque New Orleans. This is the 11th time CRN has



Dr. Rick Kaskel stops by at CRN's ASN booth and joins Terri Schleuder, Pam Woodward and Carol Hughes.



Preloaded Flash drives filled with educational materials and resources about cystinosis.

exhibited at the ASN with the goal of raising awareness and educating adult physicians about cystinosis.

Thousands of nephrologists, physicians, and researchers from all over the globe attended with the goal of learning more about kidney diseases and cutting-edge therapies to treat them. Many of those physicians stopped by CRN's booth with questions, interest, and a desire to learn more about this rare disease that is close to all our hearts in the

Cystinosis community.

This year we handed out over 100 preloaded flash drives filled with information and resources about cystinosis along with our brochures and other printed materials. Those who received the flash drives were very appreciative of the information in such a concise format. The drives included our logo and website address too. Many familiar faces in our community stopped by our booth grateful we were there spreading the word including Dr. Kaskel, Dr. Zaritski, Dr. Goodyer, Dr. Gipson and Dr. Ingelfinger among others. But we also met many new faces and had an opportunity to educate and provide a human story to this complex disease that is now increasingly reaching into the adult medical world.

As our adult cystinosis population increases we feel a continued urgency to be sure the physicians treating them have as much information as possible and know where to look for additional resources.

I know we are all familiar with the phrase, 'it is a small world'. We experienced that directly with an unexpected visitor to our booth, Dr. Ana Taveras, a pediatric nephrologist from Vero Beach, Florida, who is a neighbor to exhibitor, Carol Hughes. It was a pleasant coincidence for both of them.



Carol Hughes meets her Vero Beach neighbor, pediatric nephrologist, Dr. Ana Taveras at the ASN.

CRN would like to thank Pam Woodward, Carol Hughes, and Terri Schleuder for taking the time out of their busy lives to come to the ASN in New Orleans and help achieve our mission of educating and raising awareness about cystinosis at this venue.



Midwest Cystinosis Gathering Held in Grinnell, Iowa, August 4th-6th

By Doretta Hoffmann

The Wagner Family hosted the Midwest Cystinosis Gathering on August 4th – 6th in Grinnell, Iowa. We thank Horizon Pharma for sponsoring the gathering this year.

Friday evening we met at the local pizza place for some great food and conversation. Later in the evening we were back at the hotel to discuss the continuing research that is ongoing with cystinosis.



Summer Street Park Picture from the Grinnell's Greatest Race –

Front Row: Jon Hoffmann, Eric Gard, Mikaela Gard, Dena Gard

Back Row: Don Hoffmann, Doretta Hoffmann, Bill Croce

Saturday morning we all participated in Grinnell's Greatest Race created by Nick and Emily. The families drove to five different sites in Grinnell using a series of clues that were given to us. Once we found the correct site, we took our picture by the sign and moved to the next clue. The Hohl/ Reuter team was the first to arrive at the Wagner home and claim the prize.

Saturday afternoon and evening we gathered at the Wagner's home for some great food and some competitive games. We also decided that the Hohl family will host the 2018 gathering in Sioux City, Iowa. More details will be provided at a later date. Everyone is always welcome to our gathering.



Front Row: Nick Wagner, Emily Swim, Candy Wagner, Joan Hohl, Mikaela Gard, Eric Gard, Dena Gard, Holly Reuter

Back Row: Don Wagner, Lonnie Hohl, Bill Croce, Sarah Hoffmann, Andrew Hoffmann, Doretta Hoffmann, Jon Hoffmann, Shawn Reuter, Don Hoffmann

Not Pictured: Gabe, Aimee, Amelia, Riley Wagner and Jay & Kristy Rollingner

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Cystinosis Mexico Update: Cystagon Now Available to Patients

By Victor Gomez

On August 5th, 2016, Cystinosis Mexico Celebrated its 8th Cystinosis Symposium. We were delighted to have for the very first time Dr. Larry Greenbaum participating as a speaker who along with these great physicians, Dr. Craig Langman and Dr. Ewa Elenberg, shared important information with us like:

Cystinosis: Physiopathology, diagnosis and treatment, Endocrinology Issues, Adherence treatment and Carnitine – Nutrition in Cystinosis.



Cystinosis patients and families along with Dr. Larry Greenbaum, Dr. Craig Langman and Dr. Ewa Elenberg attend the 8th Cystinosis Symposium in Mexico City on August 5th 2016.

We would like to give special thanks to Cystinosis Research Network, and Horizon Pharma for sponsoring this meeting.

Over the last 10 years, Cystinosis Mexico has been working on having Cystagon available to Mexican patients. This has been a very hard topic, but lately in 2017, our organization, Recordati Rare Diseases, Mexican Health & government authorities have achieved this. Now Cystagon is available in Mexico, and starting January 2018, will be given by most hospitals in our city.

We are glad that this just happened! We are working hard to support cystinosis patients in Mexico.

Kevin McCalla Debuts 2nd Cystinosis Inspired Comic Book at CRN Family Conference

By Terri Schleuder

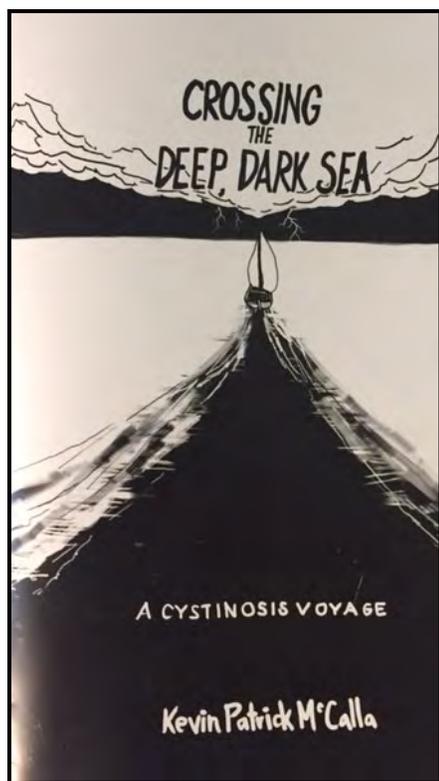
"In the depths of winter I finally learned that within me there lay an invincible summer."

-Albert Camus

Kevin's second cystinosis inspired comic book is a reflection of what living with a rare disease means to those who actually live it. His incredible artwork and thoughtful words are a metaphor that beautifully, and hauntingly capture the struggles, challenges, and finally the hope that is the life of those living with cystinosis and those who love them.

Kevin debuted his book at the CRN Family Conference in Snowbird, Utah this past July. Those in attendance received an autographed copy. If you would like to have one of these beautiful books please check out this link on CRN's website:

<https://cystinosis.org/news/announcements/242-cystinosis-comic-book-series-2017>



Graphic Artist, Kevin McCalla, signs his second cystinosis inspired comic book titled, "Crossing the Deep, Dark Sea...A Cystinosis Voyage" at the CRN Family Conference in Snowbird, Utah.

Finance Update

By Jenni Sexstone– Treasurer

Financial Review – Accrual Basis

For the nine months ended Sept 30, 2017

Revenue

For the nine months ended September 30, 2017 total income collected of \$317,000 and is significantly higher than the same period in 2016. This is attributed to additional corporate grant contributions for program services support.

Expenses

Total operating expenses of approximately \$572,000 were higher than the same period for the year prior. CRN funded \$215,000 in research grant payments compared to \$58,000 in the nine months ending in September 2016.

Education, marketing, and general operating expenses through September were approximately \$357,000 due to the 2017 family conference expenses held in July.

CRN had net operating loss of (\$253,000) for the nine months ending September 30, 2017 as compared to a loss of (\$15,000) through Sept of 2016. This is driven by higher grant payments funding research as well as family conference expenses. Generous corporate support has provided resources to continue patient advocacy and family support for 2017 and beyond to support the cystinosis community.

Cash on hand at September 30, 2017 was \$172,000 for a net decrease of (\$97,000) in the first three quarters of 2017.



Jenni, Jordan, Josie, and Jim Sexstone

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.

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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!

Black Friday. Cyber Monday

#GIVINGTUESDAY™

November 28th, 2017

#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 third year in a row, the CRN is an official #GivingTuesday partner. Leading up to and through November 28th, 2017, 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>.



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Research Update

By Christy Greeley, VP Research, Executive Director

The Cystinosis Research Network utilizes a Scientific Review Board comprised of leading experts on the disease of cystinosis which reviews grant proposals and submits funding recommendations to the Cystinosis Research Network. More specifically, the Scientific Review Board provides independent, objective review and recommendations regarding each research proposal utilizing grant review guidelines established by the Cystinosis Research Network and in accordance with the mission of the organization.

Priority is given to interventional research, both clinical and basic, that will lead to improved treatments for cystinosis. New investigators are particularly encouraged to apply. The Chairperson of the Scientific Review Board summarizes its recommendations and

presents them to the Cystinosis Research Network which then votes on each proposed project.

A major focus of the Cystinosis Research



Dave, Christy, Jack and Alex Greeley

Network continues to be a determined effort to secure a promising future for the cystinosis community through the support and funding of research grants that lead to improved treatments and ultimately a cure for cystinosis. CRN has a current research commitment of approximately \$350,000.00 and has funded over \$4 million total in research grants and fellowships. CRN funded a Cystinosis fellowship at the National Institutes of Health. CRN has funded research and education programs in the United States and many countries around the world including Egypt, Mexico, England, Scotland, Italy, Belgium, France, and much more. CRN research topics are aimed at every aspect of cystinosis with the purpose of understanding cystinosis and finding improved treatments and a cure. Topics include research and therapies related to neurological, genetic, ophthalmological, gastrointestinal, muscular, nephrology, pulmonary, skin, improved medications, psychological and much more.

CRN 2017 Call for Proposals Results in Funding for New Research Study

As a result of the 2017 Call for Proposals, CRN is pleased to announce funding for a new research project:

Study of Neuronal Structure and Function Changes in Cystinosis, John Foxe, PhD and Krishnan Padmanabhan, PhD

Department of Neuroscience Ernst J. Del Monte Institute for Neuromedicine, University of Rochester School of Medicine and Dentistry, Rochester, NY

Year One: \$110,000

Investigators at the Department of Neuroscience Ernst J. Del Monte Institute for Neuromedicine, University of Rochester School of Medicine and Dentistry Rochester, NY have outlined a research proposal taking a novel systems neuroscience strategy to address the link between molecular/cellular pathology of the lysosomal system in neurons and the resultant changes in the structure and function of neuronal circuits using new electrophysiology, imaging and computational methods to understand the effect of cystinosis on neural circuits. By identifying the mechanisms underlying the neuropathology of the disease at the basic science level, this research program will provide important biomarkers for tracking disease progression, could identify new sites/ targets for intervention and guide in the development of strategies for treatment.

In addition to this new project, CRN continues to fund extensions of the following studies:

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

Departments of Neuroscience and Pediatrics, Albert Einstein College of Medicine, Montefiore Medical Center, Bronx, NY

Year two: \$100,980

Travel Addendum: \$38,352

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 signaling as a cause of reduced adhesion and increased motility of renal epithelial cells in Cystinosis – E. Ivanova, L. van den Heuvel, E Levtchenko (Principal Investigator)

Katholieke Universiteit Leuven, Belgium

Year Two: \$88,493

Cystinosis is a genetic disease manifesting early in life (\approx 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.

Please visit the Research page on the CRN website for updates on CRN funded studies as well as other research from around the world. Also be sure to check out the many cystinosis related articles and publications available in our Article Library at <https://cystinosis.org/research/article-library/cystinosis-overview>.



August 2017

New Cystinosis Study from McGill University Montreal Children's Hospital (CTNS Nonsense Mutation Screen - CyNoMuS)

The purpose of this message is to invite cystinosis patients to join our upcoming study concerning a specific type of genetic alteration called a "Nonsense Mutation". Although cystinosis is caused by many different disruptions of the CTNS gene, we are particularly interested in "nonsense mutations" which trick the cell into stopping production of cystinosin protein. Several pharmaceutical companies are working hard to develop medications related to a well-known antibiotic (gentamicin) that permits the cell to disregard nonsense mutations.

No-Cost Genetic Testing:

Nonsense Mutations are estimated to account for about 10-15% of cases in other genetic diseases, but the prevalence in cystinosis is unknown and there is some evidence that CTNS Nonsense Mutations are clustered in certain regions. Looking forward, cystinosis patients may wish to know whether or not they carry a nonsense mutation. We would like to survey the cystinosis community and characterize the prevalence and distribution of Nonsense Mutations among cystinosis patients in North America and Europe. This would simply involve (prepaid) mailing a sample of saliva to our research group at McGill University Children's Hospital in Montreal. We would analyze the sequence of your CTNS gene and let you know for future reference whether or not you carry a nonsense mutation.

To participate in the no-cost genetic testing, please contact us by phone: (514) 412-4400 ext. 22953 or by email: murielle.akpa@muhc.mcgill.ca

Patients Who Have Already Undergone Genetic Testing

For patients who have already undergone genetic testing, you may confidentially submit your results to be included in this important study. To do so, please contact us by phone: (514) 412-4400 ext. 22953 or by email: murielle.akpa@muhc.mcgill.ca

Principal Investigator:

Paul Goodyer, M.D.
Professor of Pediatrics at McGill University
Department of Pediatric Nephrology, Montreal Children's Hospital

Study Coordinator:

Murielle M. Akpa, PhD

Education & Awareness Update

By Terri Schleuder– Vice President of Education & Awareness



Terri Schleuder

The second half of 2017 has been full for CRN. Much of the year was involved in planning and executing our 8th biennial Family Conference held in the beautiful Wasatch mountains of Snowbird, Utah. This year, we offered 'live streaming' for some of the research presentations allowing many who could not be there in person to still be a part of this great conference. A huge debt of gratitude goes to our fantastic co-chairs, Pam Woodward and Christy Greeley for their leadership and hard work to make the 2017 CRN Family Conference so successful.

There were many highlights. Two particularly exciting ones fit very well with this year's theme "Soaring to New Heights". Dr. Stephanie Cherqui, presented her research in stem cell gene therapy that could potentially lead to a cure for cystinosis, and Dr. Jennifer Simpson, discussed her research using of the cysteamine nanowafer to treat corneal cystinosis. The future of improved therapies seems brighter than ever.



Carl and Steve Schleuder

In August, CRN presented two \$1,000 academic scholarships to two very deserving young people. Emily Patterson was the recipient of the CRN Academic Scholarship for an Individual with Cystinosis and the Sierra Woodward Academic Scholarship for the Sibling of an Individual with Cystinosis was presented to Tyler Morrill. (See pages 4 and 5 page for more about these two and their academic plans.)

2017 also saw some changes to CRN's Board of Directors. After serving for the past five years, and doing an excellent job leading our organization, Jeff Larimore stepped down as President. We are so grateful for his vision, and leadership that continues to shape CRN as a premier cystinosis support organization. Jeff will continue to serve as a board member. Clinton Moore has stepped into the role of President and we look forward to his energy, charisma, and new ideas to keep CRN moving forward as the great organization it is.

Current board member Ina Gardener assumed the role Secretary. Former V.P. of Development, Jen Wyman, has returned as the V.P. of Family Support. Jen and her family have been active in, and supportive of CRN for many years.

We also welcome three new board members, Aimee Adelman, Jill Emerson, and Larry

Thomas. We look forward to their energy and enthusiasm. Please read more about our new board members on pages 6 and 7.

In November, CRN exhibited at the 2017 American Society of Nephrology conference (ASN) held in New Orleans this year. Pam Woodward, Carol Hughes, and Terri Schleuder continued the tradition of educating adult nephrologists in attendance about cystinosis, so they can more adequately meet the needs of your growing Cystinosis adult population. (see pages 28 and 29)

We are also in the beginning stages of planning to send several representatives from the cystinosis community to the 2018 Rare Disease Week on Capitol Hill. Organized by the nonprofit, Every Life Foundation, this event will occur in late February, coinciding with Rare Disease Day, whose theme next year will be as it was in 2017, Research. There is no better time in recent memory to make our legislators aware of the needs of the rare disease community as they consider and pass legislation that will impact us all.



Eric, Steve and Chris Schleuder try to beat the heat, in Orlando, Florida.

Rare Disease Day 2018 Theme Announced

The eleventh annual Rare Disease Day will be celebrated on February 28, 2018. The theme this year is the same as last year and focuses on **Research**. Research brings hope to people living with a rare a 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 2018 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

When I was asked by Jeff Larimore to take on the Vice President of Development responsibilities on an interim basis I thought to myself, "Self, be careful not to take on more than you can handle. Do not go outside your comfort zone." I also knew there was a high probability the interim designation would transition to a permanent designation. Well, I agreed, and the interim designation became permanent. I am now in my second term. I have to admit, my comfort level in the beginning was not high. I decided to accept the position because of the opportunities to grow and the need was real. Now, some folks would think of this as being selfless (others perhaps not so much). For those who are wondering, I took on this role to further our cause. "If not me, who? If not now, when?"



The Morales family, Christian, José, Velyna and Alexandra

After five years of performing these responsibilities, I continue to be humbled by the actions and level of commitment of our community. I have learned a lot since day one. One of the major questions I had to work through was: Why do people donate their money? The number one reason I learned is that the donor receives a tax write-off. Although rule of thumb says someone receives a net financial gain from donating, this is rarely the case. Giving usually cost money, you give to give and not to get. People are not giving money with the expectation of making a profit; clearly there are other reasons for philanthropy. The following are six plausible reasons related to philanthropy:

IMPACT – one key reason people give is that they want to make a positive difference in the world. Saving lives, reducing suffering, feeding the hungry, eliminating disease are examples of what is at the heart of giving. Contributors do not expect anything in return, what they do expect is tangible evidence that their contribution is for the good.

APPRECIATION – Confirmation of an act. People and organizations donate with a the expectation their action will be recognized on an individual or organizational basis; i.e., confirming appreciation for something that was done for them or someone they love or, simply from gratitude for the position impact an organization is making.

MISSION – when individuals are vested in a shared interest of an organization, they give because they endorse and share in the organization’s vision and mission. Better said, these individuals share a common purpose and have a high sense of commitment toward achieving the targeted results.

IMPULSE – an emotional effect can also impact gifting. Personalized because you experience the need on a first hand basis. You witness the extreme nature of the pain and suffering and even though there are other organizations with equally valid needs, you decide to support a particular organization because of the personal connection.

RECOGNITION – in some instances, all an individual or company is seeking is the attention that comes from donating. This can come in different forms, personal, private or public recognition, naming of an event in your honor, receipt of an award. My West Point class, Class of 1978, recently donated \$1.0 Million for naming rights for the plaza in front of the new West Point Visitor Center.

BENEFIT – a more controversial motivation for donating to a cause is for personal benefit. Something to be received in the form of a tangible benefit for a portion of the contribution made. In effect these are two transactions: one is receiving services or goods and the second is the act of donating.

So, the question is what motivates you? What do you want out of donating? Whatever it is, please remember, "If not me, who? If not now, when?"

If you are inspired to support CRN and assist in pursuing a cure for cystinosis, please contact me via email or mobile: jose.morales01@icloud.com, 203-722-9292.

We remain committed to all of you, on an individual and collective basis, and will continue to take action and drive change for the benefit of our children.

José T. Morales
Vice President - Development



José and Christian Morales at the NYC marathon.

Federal Drug Administration (FDA) - Center For Drug Education and Research (CDER) Workshop for Effective Advocacy in Rare Diseases Drug Development

By José T. Morales



CRN's VP of Development José Morales, and Hoizons's Senior Director of Global Patient Advocacy, Tricia Mullins, attend the FDA Workshop on Effective Advocacy in Rare Diseases Drug Development.

The Food and Drug Administration is a federal agency of the United States Department of Health and Human Services, which is one of the United States federal executive departments. The FDA is responsible for protecting the public health by ensuring the safety, efficacy, and security of human and veterinary drugs, biological products, and medical devices; and by ensuring the safety of our nation's food supply, cosmetics, and any products that emit radiation. The FDA is responsible for advancing the public health.

As part of the U.S. Food and Drug Administration (FDA), The Center for Drug Evaluation and Research (CDER) regulates over-the-counter and prescription drugs, including biological therapeutics and generic drugs. The CDER performs an essential health task for the public: confirming safe and effective drugs are available for people in the United States, so they can improve their health.

The recently completed workshop provided valuable information on how to engage with CDER and better understand the beneficial overlap of effective advocacy techniques and FDA regulations in rare disease development. The workshop consisted of an extensive agenda:

What is the FDA and Who is Involved with Rare Diseases Engagement?

- FDA Orphan Medical Product Designation Program
- CDER Divisions Working with Rare Diseases
- Professional Affairs and Stakeholder Engagement within CDER

Types of Patient Engagement with CDER at FDA

- CDER Patient Engagement and Interactions
- Externally – led Patient – Focused Drug Development Meetings
- Patient Focused Drug Development (PFDD) Meetings with FDA

Developing an Effective CDER Engagement Strategy

- CDER Centric Best Practices
- Patient Advocate Centric Best Practice

Determining and Developing Next Steps

One of the remarkable developments of recent history in rare disease research has been the evolving role of patient advocacy groups and the collaborative partnership among such groups and the scientists who study rare diseases, as well as the government officials charged with overseeing medical research and regulatory processes. This collaboration is unparalleled in other areas of medical research and product development. It has emerged as a major change agent impacting the adoption of public policies, available research funding and other factors affecting the general climate for research on rare diseases.

The Cystinosis Research Network is committed to addressing the entire spectrum of the community's needs. It is through participation in these types of workshops and events that we continue to ensure the Voice of the Community is heard.

Corvette Club of Western MA and Jenn Loglisci Fundraise for CRN

By Jenn Loglisci

The Corvette Club of Western Mass., Inc. got together with me on Saturday September 16th, 2017 at Cindy's Drive In in Granby, MA to raise money and awareness for the Cystinosis Research Network. Corvette Club members from all over Western MA attended. Prior to the event flyers were placed around Western MA and information was shared on social media. Visitors of this event enjoyed food from Cindy's while looking at all the cars. I set up a table with pamphlets, the cystinosis comic books, the 2016 art calendar and past CRN newsletters. People were very interested in



hearing about cystinosis, a disease they never heard of from a person living with the disease. The weather was

beautiful and the DJ set the classic car mood with 50's music while sporadically announcing the purpose for the event. A raffle table was set up with prizes to a corvette dealership, car cleaning product basket, health therapy bags, gift cards and much more.

We have already booked the event again for August 18th, 2018 at the same location.

This event would not have been possible without the president of the club, my Uncle Rene Lajeunesse who helped me put this event together.





Chandler Moore

Chandler's Chance 5th Annual Christmas Palooza For Cystinosis

In Honor of Chandler Moore

Benefiting: Cystinosis Research Network

Saturday December 2nd 2017

5:30pm until 9pm

Address: 24062 Peterkins RD Georgetown Delaware 19947

All Activities are FREE of charge although DONATIONS are
APPRECIATED

Please make checks payable to: Cystinosis Research Network or donate
online at www.cystinosis.org

Hay Rides, Fire Truck Rides, Face Painting, Make Reindeer
Food, BB Gun Shooting Range, Photo Booth, Mr. and Mrs.
Santa Claus, Coffee, Hot Chocolate, Cookies, Popcorn,
Cotton Candy, Food Trucks, Bring Chairs and Blankets
LIVE ENTERTAINMENT BY THE DIRT ROAD OUTLAWZ!!!



SPONSORED BY: HORIZON & LEADIANT PHARMACEUTICALS



Chandler's Chance is always a fundraising highlight for CRN. Thank you to the Moore family and all those involved each year that make this such an outstanding event.

For the last three years it has coincided with Nemours Family Cystinosis Day. Since both events occur after we have gone to press look in our next Cystinosis Advocate in June for highlights.



SAVE THE DATE!

3rd Annual Nemours Family Cystinosis Day ~ **Saturday, December 2, 2017 10:00am – 3:30pm**

We invite patients and families to this fun-filled and educational conference aimed at providing you with the latest updates in the care of nephropathic cystinosis. This year, we are happy to have Dr. Timothy Bunchman, Pediatric Nephrologist of Children's Hospital of Richmond at VCU, join us to discuss current advancements in cystinosis!

Location: The Clubhouse at Baywood, 32267 Clubhouse Way, Long Neck, Delaware, 19966

Travel and Lodging: We have applied for a grant, approval pending, which will cover a portion of travel and lodging for qualified attendees

Scheduled Speakers:

Dr. Joshua J. Zaritsky, *Division Chief, Pediatric Nephrology - Nemours A.I. DuPont*

Dr. Timothy Bunchman, *Professor and Chief, Pediatric Nephrology - Children's Hospital of Richmond at VCU*

Dr. Mihir Thacker, *Pediatric Orthopedic Surgery - Nemours A.I. DuPont; Cystinosis Research Network Medical Advisory Board*

Following the conference, you are invited to attend the **5th Annual Chandler's Chance...A Christmaspalooza for Cystinosis fundraiser from 5:30PM - 9:00PM** at the Moore family residence, 24062 Peterkins Road, Georgetown DE 199470

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.

Jen Wyman
VP Family Support, Cystinosis Research Network



Jan Julian and Jen Wyman at the 2017 CRN Family Conference



Join the Cystinosis Research Network Today!

Immediate Family	\$20.00
Extended Family / Friend	\$25.00
Professional	\$35.00

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

Payable in US dollars

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

Name _____

Street _____

City & State _____ Zip Code _____ Country _____

Phone _____ Fax _____ Email _____

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.

Support CRN's Mission with Your Donation

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

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

Name _____

Street _____

City & State _____ Zip Code _____

Phone _____ Fax _____ Email _____

In Honor Of _____

In Memory Of _____

You may send notification of my gift to:

Please check all that apply:

- Friend
- Parent of Child with Cystinosis
- Family
- Individual with Cystinosis
- Professional
- I am interested in volunteering for CRN. Please contact me.



Bree Forrester

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, Chicco, Coldwater Creek, American Eagle Outfitters, and many more!





United Way Contribution Guidelines

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

Agency Name	The Cystinosis Research Network, Inc.
Non-Profit Tax ID #	04-3323789
Address	302 Whytegate Ct., Lake Forest, IL 60045
Telephone	1-866-276-3669 (toll free), 1-847-735-0471
Fax	847-235-2773
E-mail Address	CRN@cystinosis.org
Web Page	www.cystinosis.org
<p>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.</p> <p>The Cystinosis Research Network prepares all necessary documentation and submits it to the respective local United Way organization.</p> <p>The local United Way organization processes the documentation and sends a check for the aggregate sum designated for the Cystinosis Research Network.</p> <p>The Cystinosis Research Network sends thank you/acknowledgement letters to recognize contributing individuals.</p>	

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:

https://org.amazon.com/npo/portal/marketing-tools/ref=org_prt_gwh_mt

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



Dedicated to a Cure. Committed to our Community.

Cystinosis is a rare, genetic, metabolic disease that causes an amino acid, cysteine, to accumulate in various organs of the body, including the kidneys, eyes, liver, 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.

302 Whytegate Ct.
Lake Forest, IL 60045
www.cystinosis.org

Phone: 847-735-0471
Toll Free: 866-276-3669
Fax: 847-235-2773
Email: info@cystinosis.org

Editor: Terri Schleuder

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.*

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