Ring in the Future: 2019 Family Conference in Philadelphia

The Cystinosis Research Network is pleased to announce that its 2019 Family Conference will be held July 18 – 20, 2019 at the Hilton Philadelphia at Penn’s Landing. Please plan to join us to reconnect and learn more about new research findings, receive updates from cystinosis organizations from around the world, meet and renew friendships with other families, have the opportunity to participate in research studies, and interact one on one with many of the world expert clinicians treating and researching cystinosis.

The Hilton Philadelphia at Penn’s Landing is a newly renovated property, the only hotel on Philadelphia’s beautiful waterfront in the historic district. They offer an indoor pool, lobby bar, 24-hour coffee shop and waterfront dining at Keating’s Rope & Anchor, Bar plus...
Since the last newsletter the Cystinosis Research Network has been buzzing. We currently have so many things going on, at times it’s hard to keep up with it all. I am so proud to be part of such a wonderful, hardworking organization with so many other extremely hard-working individuals. It’s almost overwhelming thinking back to when my son was first diagnosed and how the Cystinosis Research Network reached out and assisted me and my family. I often hear the same thing from other families as well.

I can remember avoiding all things cystinosis related during the first eight years of my journey. I knew this organization existed, as I would receive this exact newsletter twice a year. I would flip through it and see all the wonderful families doing so much for this organization and for the cystinosis community in general. I can remember when I first started getting involved just a few years ago. I was truly honored when I was asked to be part of the Cystinosis Research Network as a board member.

CRN continues to give excellent support to families, exactly like they gave to me and my family years ago. If you ever have a need or concern for anything cystinosis or non-cystinosis related please reach out to us. We have many resources and we are here to help. This is your advocacy group.

CRN also continues its commitment to fundraising, supporting research, educating, and raising awareness. Many recent efforts are included in this newsletter. Along with many other important areas of interest to the rare disease community.

Again if you have any interest in being part of these efforts, please reach out to us. There is always something to do. There is always awareness to raise and it’s easier than you think. There are many different ways to get involved.

So, for now and always...We will continue to support this community in every way possible. We will grow, strengthen relationships, fight for what this community needs and deserves, and continue to do whatever we can to improve the lives and health of all of our cystinosis community members.

Sincerely,
Clinton Moore.
UCSD Cystinosis Research Study Seeks Adult Participants

Dr. Doris Trauner and her research group at the UCSD School of Medicine are conducting a research study to evaluate the relationship between sleep, memory and thinking in adults with cystinosis. We are looking for adults with cystinosis to participate in an overnight sleep study and cognitive testing focusing on memory.

Participation involves completing questionnaires, performing tests of memory and thinking, and sleeping overnight in a UCSD sleep laboratory. The questionnaires and memory and thinking assessments will take about 5-6 hours to complete. The sleep study portion will require an overnight stay in a sleep lab with the placement of electrodes for recording bodily functions such as heart rate and muscle movements, but will be completely non-invasive and non-painful. The study will pay for travel and meals, and participants will receive the results of both their memory and thinking tests and sleep assessments. If any problem is found in the sleep study, we will send the results to your primary care physician.

For more information, please contact Jennifer Crowhurst at jcrowhurst@ucsd.edu or Tammy Vu at 858-822-6700.

Support CRN through Cafepress

https://www.cafepress.com/cystinosis2018

Support CRN, raise cystinosis awareness, and purchase some fine merchandise all at the link above. It is absolutely a Win-Win.
The Cystinosis Research Network is pleased to announce we will be accepting applications for our two $1000 academic scholarships until August 15, 2018. One is an Academic Scholarship for an individual with cystinosis and the other is the Sierra Woodward Sibling Scholarship for the sibling of an individual with cystinosis.

Information about both Scholarships and the application forms can be accessed on CRN’s website:

https://cystinosis.org/family-support/scholarships

Both Scholarships are available to persons attending a qualifying College, University or Trade School. They are offered to those beginning their college career or already attending college. They may also be reapplied for in subsequent years.

Completed applications should be received at the address below by August 15, 2018 to be considered.

CRN Scholarships
C/O Terri Schleuder
40472 Franklin Mill St.
Novi, MI 48375

Tyler Morrill, winner of the 2017 CRN Sierra Woodward Sibling of an individual with cystinosis academic scholarship.

Emily Patterson, winner of the 2017 CRN Individual with Cystinosis academic scholarship.
CRN Family Conference (cont. from page 1)


Registration materials will be available in early 2019 on the CRN website and will also be mailed to families. Special room rates have been negotiated with the hotel and will be made available to conference attendees.

We look forward to seeing you in Philadelphia in July 2019.

2017 CRN Family Conference attendees from Snowbird, Utah.

Please plan to join us in 2019 in Philadelphia to connect and reconnect with new and old friends from the Cystinosis Community. See you there!

The Cystinosis Research Network proudly debuts our new look. Check our logo (right) and visit us at: cystinosis.org to see CRN’s updated website.

I love that the new logo shows both the closeness of the cystinosis community & what really matters...the people, the patients, parents, caregivers, and all those involved with us in this journey.

-Clinton
Rare Disease Week on Capitol Hill 2018

By Aimee Adelmann and Katie Morrison

Rare Disease week is an annual event in Washington, DC that brings together those who personally experience and those whose lives have been touched by someone with a rare disease to learn about legislative advocacy first hand. The objectives are to bring education and awareness to federal legislators and to advocate for positive and meaningful change that will ultimately better the lives of those with rare diseases in our country. We are 7000 diseases strong! It is inspiring and empowering to see everyone together. Standing with people who were attending for the first time along with seasoned advocates brought me new and different perspectives and a lot of energy!

I had the opportunity to see how many initiatives had been successfully implemented and to learn about new efforts for the future. It is truly powerful to see that sharing our real-life experiences and asking for what we need can really create positive changes.

Our Cystinosis community was well represented this year. We had 16 people participate. One of those individuals was a Future by Design board member, Katie Morrison. Katie’s insights and experiences follow:
When arriving in Washington D.C. for Rare Disease Week I was most excited about visiting D.C. for my first time without an appointment at the National Institute of Health, as many cystinosis patients can attest to. The second thing I was very excited about was to participate in rare disease advocacy. I have volunteered in the past for the National Kidney Foundation, Donate Life, and of course Cystinosis Research Network. But this was my first time going "behind the scenes" to see what it takes to be involved and heard on a state and federal level. I learned there are 7,000 rare diseases but only 300 have treatments. If you do the math, that equals about 95% that have no FDA treatment. With numbers like that the cystinosis community is pretty lucky! I’m very fortunate and grateful – that is the way I think of it. At Rare Disease Week on Capitol Hill I met many others from the rare disease community, and although our diagnoses are very different, we all have the same concerns and needs. There are still many who need treatments and a cure.

As mentioned before, there are 7,000 rare diseases and very few with treatment. One of the pieces of legislation that I learned about is the Open Act. This legislation could bring safe and effective treatments to those with rare diseases. The Act also provides incentives for pharmaceutical companies to develop drugs to treat these rare diseases. This is bipartisan legislation that is supported by over 220 patient organizations, including Cystinosis Research Network and the National Organization of Rare Diseases (NORD). This Act correlates strongly with the Orphan Drug Act, which is a law passed in the United States in 1983 to facilitate drug development for rare diseases. Meeting so many others affected by rare disease was a constant reminder of how fortunate the cystinosis community is to have FDA-approved treatments. It was also a constant reminder and motivation to stay involved. As a community we are small, but the old adage goes, 'there's strength in numbers'. We have worked so hard to spread the word of cystinosis and create a community to support patients and care givers. We have so much as a community and are so fortunate. Many others affected by rare disease do not even have treatment.
Being able to speak with my state’s legislators was a great way to spread awareness for cystinosis and others with rare diseases. Cystinosis is a great example of what drug development can do for those living with a rare disease. I hope my story inspired them to support the Open Act so others can have treatment. In order for this, and other bills to pass there needs to be support. You can support this bill by emailing your state’s senators and representatives and asking them to support the Open Act and the Cystinosis community. Rome was not built in a day, but with patience and perseverance your voice does matter and something as simple as sending an email to show support can help others. Let’s give back to the rare disease community, after all, we all want one thing... safe and effective treatments for full and happy lives.

Katie and I invite you all to get involved, contact your federal representatives and senators, and learn about different legislative efforts that might affect you. Your voice does matter and can make a difference. With our efforts on Capitol Hill and the support of Delaware Senator Christopher Coons, our community was even able to create Cystinosis Awareness Day on May 7th!
The 3rd Annual Nemours Cystinosis Family Day

By Clinton Moore

The 3rd annual Nemours Cystinosis Family Day is in the books and was the best one yet. Since this event is growing at a very rapid pace, we had to move it to a larger venue already in just the 3rd year. This year it was held at the Baywood Golf and Country Club in Long Neck, Delaware.

The coordination and planning of this event is a combined effort from Jill Emerson, Dr Zaritsky, and myself.

This year we had our highest attendance numbers ever, and it was our pleasure to welcome 16 cystinosis families as well as other industry partners and physicians.

The agenda ranged from talks about the general overview of cystinosis, to transplants, to diets. There was even some time for casual conversation and mingling among attendees during lunch and other breaks.

This year we were able offer cystinosis families a stipend for their lodging and reimburse their travel expenses. Our effort was to make this a ‘no out of pocket’ expense event for the families. This event is also held on the same day as the Chandlers Chance Christmas Fundraiser, so they got to enjoy that as well.

We hope that everyone that attended had a great time and is looking forward to attending again this December. The event is promised to be educational as well as amusing. Details for the next event will be available soon.

Attendees of the recent 3rd annual Nemours Cystinosis Family Day brought 16 cystinosis families together along with physicians, and other industry partners to learn about cystinosis, connect with each other and have fun. It was a great success.
By Clinton Moore
This 57-mile walk was inspired by a movie called the Ataxian. I watched it during Rare Disease Week in Washington D.C. back in February. While watching the film, my mind drifted to thinking about what could I do on the extreme side for the cystinosis community. Two other walkers pledged to endure the entire 57 miles with me. Both good friends....Cody Collins and Elaine Hyler. Other people will be joining for a part of the journey too. We feel good....we are ready. It’s time to walk all over cystinosis.

11:40 AM We are off to a late start, but the gang is all here, and we are ready to go. The current plan...To pound as many miles as we can...as fast as we can.

2:00 PM Before beginning the walk, I agreed to complete some “dares” during the race. Kind (or perhaps cruel) donors, provided CRN with $50 donations to see the dares come to life. A little while ago I got a taste of the first dare – dressing like a baby – I wore a diaper and sucked a pacifier for an entire mile! I may be starting to regret this part of the commitment.

4:00 PM It looks like we are averaging three miles an hour which should put us at 57 miles around noon tomorrow.

6:00 PM We are over 15 miles in, and Annie and Chandler have informed me there is a coconut bra and grass skirt with my name on it. This should be cute. Overall everyone is holding up good so far. Spirits are high.

8:00 PM Just got word that burgers are waiting for us at our next checkpoint.....which
will be great. There is a 3-legged dare just before sunset. At this last rest stop people started bending over and grabbing their knees to rest – perhaps fatigue is setting in? Also, we just got word that downpours are imminent. Along with almost guaranteed thunderstorms.

**10:00 PM** We are now under a tornado watch until 1 am. At around 10:30 pm we had to take cover under a pavilion, but the rain will not dampen our efforts. This storm is severe. Heavy lightning and thunder, downpours, and high winds. We found a large enough area to continue walking after moving a dozen or so picnic tables. There are a few other people also taking cover under the pavilion. It looks like they are playing a card game of some sort. We are doing our best not to get soaked as temperatures are plummeting.

**1:00 AM** The pavilion was our safe place for the last three hours. However, it is getting late, everyone looks and feels exhausted and those small circles and constant left turns are enough to drive anyone bonkers. So we moved on. Nate joined the walk.

It is now 60 degrees! Who’s making fun of my jeans now?

**3:00 AM** Cody has just informed me he cannot go on. He gave it 100% effort. He’s been struggling for the past couple hours with foot pain.

**3:30 AM** Elaine just told me she must lay down. She cannot go on either. She is also experiencing foot pain. I have Nate and his legs are still fresh.

**4:30 AM** The internal struggle begins. The thoughts of... ‘I can’t do it, and I won’t do it again’ are haunting me. Should I make a change in trail? This road is beating me up. The large rocks are terrible on my feet. I just stare at the road. It looks no different than the sky. Dark. All outside communication has stopped. No texts. No calls. No Facebook notifications. It’s quiet. Too quiet. Everyone is asleep as they should be. My own mental demons are telling me to quit. I feel defeated and alone. I am exhausted. I grabbed some snacks in hopes of an energy boost, but it doesn’t seem to be working. I’m pretty sure I just swallowed a bug. Even Nate and I are barely talking. The mental fatigue at this point is outweighing the physical by far.
6:00 AM The sun is back and it’s like a breath of fresh air. We made a trail change to ease the walking efforts. There is storm damage everywhere.

8:00 AM Less than 13 miles to go... Just got a text from Clair. She has coffee and a bagel waiting for us.

11:00 AM Here we go – a rare dare to make a rap song to include Elijah (Emily Mello’s son). Our group came up with it together and I was happy to nail it on the first take! We wanted to be finished during this hour. Watching those minutes tick by, and having the finish line beyond my viewpoint are a blow, but we keep moving.

1:00 PM There are 1.7 miles left. The original plan had us finishing at the end of this loop, but staying true to ourselves, we were a bit short due to some of the improvisations we had to make.
1:30 PM Our 57-mile mission is complete! WOW – the emotions! Family and friends met us at the end and this journey and have left each of us with tears in our eyes.

This was beyond tougher than I thought. I completed over 400 miles of training walks that helped....but the mental stress was extreme. That being said, I would gladly endure this again as well as much more for this community.

You can check out videos from the journey on our Facebook page: [facebook.com/CystinosisResearch](http://facebook.com/CystinosisResearch) We are currently putting together a task force for the 2019 Cystinosis Awareness Day and determining how to expand on the success of 57 Miles for Cystinosis. Email me if you would like to be a part of these events at clintonmoore1@aol.com.
Healy Sisters Raise Cystinosis Awareness for Rare Disease Day in Minnesota

By Sara Healy

My twin sister Jana and I went to a Rare Disease Day event on February 23rd at the University of Minnesota. The goal was to bring researchers and rare disease patient populations together. It was exciting to see all the people interested in helping others who suffer from a rare disease and to see how far science has come in finding treatments. We went to this event to spread awareness of our rare disease “Cystinosis”.

We had several people visit our booth and we gave them information explaining how our rare disease affects the body. There was a girl who stopped by our booth along with her dad. She told us she was writing a paper for school on rare genetic disorders and lysosomal storage disorders, because her sibling had passed away from one. We described our lysosomal storage disorder in detail and gave her a flash drive and some pamphlets.

We also met with Evelyn S. Redtree who is a study coordinator for Lysosomal Disease Network, an organization that studies all the lysosomal disorders. She sent us an email on how to join the Lysosomal Disease Network emailing list where we could get new info and updates on these diseases, so we signed up.

We are glad we participated in this event and we hope to do it again next year. We plan to keep spreading awareness of rare diseases, so more people can understand what we really go through each day.

Sara and Jana Healy raising cystinosis awareness at a Rare Disease Day event at the University of Minnesota on February 23rd.
Meschke Family begins the Cystinosis Journey with Son, Jaxon’s Diagnosis

“Hi, We’re the Meschke’s. Brian, Chelsea, our wonderful baby boy Jaxon, and Jaxon’s giant, drooling, four legged brother Rogan. When Jax was 4 months old we noticed that he had started to fall behind on his weight gain. After a routine check up, our pediatrician decided it was time to look further into why Jax had fallen below the 3rd percentile in height and weight. Thank goodness for that! He noticed uncommon levels of glucose and protein in his urine and recognized right away this needed further attention from a specialist. That’s when we met our amazing team of doctors at C. S. Mott Children’s Hospital in Ann Arbor Michigan. In the beginning of January it was confirmed that Jaxon had cystinosis. As with any diagnosis we were crushed. In a matter of a moment our perfect little boy was going to have a much different life than we had dreamed of. With amazing support from family and friends we have found a “new normal” to life and have our sights set on those dreams and so many more for our little boy. We are a happy family of four and ready for anything that comes our way.”

Follow Us On Social Media

Don’t miss out! Stay updated on the latest support programs, educational materials, research, and events from the CRN. It only takes a moment. Follow us today!
Josie Sexstone and Family Share RDW Story with Local Media

Health Ambassador, Ben Logan girl, family share story of living with a rare disease on Capitol Hill

Benjamin Logan Elementary student Josie Sexstone spent her spring break last week just enjoying some of the perks of being a 10-year-old. She stayed up late during sleepovers with her cousin and sister and had a memorable outing to the trampoline park Sky Zone, where she jumped around until she was “red in the face,” she says.

Her family is most thankful for these somewhat ordinary and also lighthearted days for their fourth-grade, sweet-spirited daughter, who lives with a rare metabolic disease called cystinosis, which affects approximately 500 people in the U.S. and about 2,000 people in the world.

Every day, it takes an extraordinary effort from both Josie and her parents to take and administer a number of medications around the clock, with doses that could nearly fill an entire table, to treat this condition.

In addition, she also uses special prescription eye drops called Cystaran multiple times during the day to dissolve corneal cystine crystals in her eyes and relieve photophobia, pain or discomfort in the eyes due to light exposure.

While some of her medications can really taste foul, even after being mixed with juice, and also have to be taken at overnight hours – 11 p.m. and 5 a.m. – to meet the four required doses in 24 hours, Josie takes them in stride and is not one to complain. The mature-beyond her years pre-teen knows how vital these medicines are to keeping her healthy.

“It’s just something I have to do,” she said nonchalantly last week at her Zanesfield area home that she shares with her parents, Jim and Jenni, and 14-year-old sister, Jordan.

“I’m used to it; it’s part of our routine. It might not be fun, but if I just take it quickly, then it’s done."

Capitol Hill opportunity

Josie was diagnosed at age 14 months with cystinosis, which causes an amino acid, cystine, to accumulate in various organs of the body. Cystine crystals accumulate in the kidneys, eyes, liver, muscles, pancreas, brain and white blood cells, according to the Cystinosis Research Network’s website, www.cystinosis.org. Without specific treatment, including the many medications that Josie takes, children with cystinosis develop end-stage kidney failure at approximately age 9, the CRN reports.
Over the years, the progressive condition also can cause complications in other organs of the body, including muscle wasting, difficulty swallowing, diabetes and hypothyroidism.

To share some of Josie’s unique story and help advocate for others with rare conditions around the country, the 10-year-old and her family were invited to Washington, D.C., for Rare Disease Week at the end of February and first part of March. They were extended the invitation as result of their involvement with the Cystinosis Research Network, including Mrs. Sexstone’s work as a board member for the organization.

Throughout their time on Capitol Hill, the family met with representatives from the offices of Senator Sherrod Brown, D-Ohio, Senator Rob Portman, R-Ohio, and Representative Steve Stivers, R-Ohio. They also spoke with Representative Jim Jordan, R-Ohio, “who went out of his way to meet with us,” said Mr. Sextone.

Josie’s mother related how they want to use their experiences to help others with various rare diseases to also have access to treatment and specialty medications. One of the main treatment drugs that Josie takes, Cystagon, has been approved by the Food and Drug Administration since 1994 for the standard treatment of cystinosis. It is a cystine depleting agent, meaning that it lowers cystine levels within the cells. It has proven effective in delaying or preventing renal failure, and also improves growth of children with cystinosis, according to the CRN’s website.

“While this medicine doesn’t taste good and is unpleasant to take, Josie know how fortunate she is to be able to have it as it keeps her alive,” Mrs. Sexstone said. “We’ve come to realize how fortunate we are that her condition is one that has a treatment, as only 5 percent of rare diseases have a treatment available. Josie wants everyone with a rare disease to have the same access to the medication she does.”

Along those lines, the Zanesfield family said while in D.C., they were advocating for the Orphan Product Extensions Now Accelerating Cures and Treatments Act of 2017, introduced by Senator Orrin Hatch, R-Utah, which has been endorsed by more than 200 patient organizations, including the CRN, according to Rare Disease Report’s website, www.raredr.com. The OPEN ACT would provide incentives to pharmaceutical companies to
repurpose already approved drugs for the treatment of rare diseases. The main incentive would be an additional six months of market exclusivity to drugs that are repurposed and approved by the U.S. Food and Drug Administration to treat rare diseases.

This law would apply to currently approved drugs that are still under patent. The 6-month extension would be in addition to other types of exclusivity, such as pediatric or qualified infectious disease product exclusivity, according to Rare Disease Report.

**Early years and a quick diagnosis**

Long before they were serving as health advocates on Capitol Hill, the Sexstones related that the first months of their youngest daughter’s life went smoothly and without any major concerns. However, during check-ups with her pediatrician between ages six to nine months, they noticed that she hadn’t grown any taller, and at her one-year checkup, the parents and their doctor were alarmed that Josie had lost weight. “She had been throwing up some, and our doctor thought at first that she had a virus,” Mrs. Sexstone said. “But Jenni didn’t take ‘no’ for an answer,” her husband said. “We just had the feeling that something wasn’t right.” Their pediatrician ordered blood work and a urine sample for their 12-month-old girl. In the meantime, the family went on a vacation to Kelley’s Island and that’s when they received some urgent news. “We got a call while on our trip that Josie likely has some kind of kidney disorder,” Mr. Sexstone said. “They wanted us to see a doctor at Cincinnati Children’s Hospital right away.”

The former Cincinnati area residents said they saw the nephrologist shortly thereafter, who also observed that Josie had rickets, as she was bow-legged and her bones were very soft. Although he didn’t mention it to the family right away, the experienced doctor suspected that Josie had cystinosis, and sent her blood samples off to San Diego to test for that condition. In about two to three weeks’ time, the family would learn that their daughter had this very rare disease. “The news at the time, of course, was devastating to us,” Mrs. Sexstone said. “While we very much grieved hearing about this condition, we also were grateful to know what was causing these issues for our daughter. Also, it typically can take seven years to receive a diagnosis for a rare disease. It’s remarkable that we were able to connect with the nephrologist who was familiar with cystinosis and just had a feeling about Josie, so we got this figured out in a matter of weeks.”

**Our normal**

For the family of four, life these days is filled with some of the usual hustle and bustle of having a 10-year-old daughter and 14-year-old daughter, along with the medication demands and other requirements to keep Josie healthy with cystinosis. Several times throughout the year, she has appointments with specialists, including nephrology, orthopedics, ophthalmology and endocrinology. The preteen also has blood work performed four to six times a year to check on her levels. During her school day at BLES, her teachers also know that Josie needs open access to water at all times. In gym class and recess, she is encouraged to participate in any and all activities, but just
has to make sure she doesn’t get dehydrated and she takes extra precautions when playing out in the sun. “It’s our normal; we all have to adjust to make it work,” said Mr. Sexstone, who serves as the executive director of the Hilliker Y and YMCA Camp Willson. “We couldn’t do it without our family, friends and Josie’s school; they’ve all been so supportive,” said Mrs. Sexstone, who serves as a finance manager for a Cincinnati-based publications company and works remotely from her home. “The school and her teachers have been incredible. She has to take her medicine throughout the day, and they are so good about making sure she has water and anything else when she needs it.”

Older sister Jordan also provides a close-knit support for her young sibling, watching the clock and noting when it’s time for Josie’s medications, and also helping her sister to grab a snack that she needs before taking different prescriptions. “Jordan takes very good care of her little sister; their relationship is a special one,” their father said.

Another important group in the Sexstones’ lives is the Cystinosis Research Network, where the family has found overwhelming support from others living with the same condition, along with medical experts related to cystinosis. They typically attend conferences with the organization every other year in locations around the country. Josie named a number of friends she has made through the organization, other children with whom she shares a close bond and who deal with the same challenges of this genetic condition. “This is the place where I feel like I belong,” she said of the conferences. “My friends there, they know what I go through. One of my friends, Casey, even taught me how to do the eye drops.”

The BLES student’s current good health and activity levels, including one of her favorite activities, playing basketball, are certainly not taken for granted by her and her family. Even with the specialized prescription medication treatment, individuals with cystinosis typically need a kidney transplant sometime in their teen years, but that age can vary at times.

Other interventions that the 10-year-old might eventually need include knee surgery and a procedure on the arches in her feet, her parents said. But with her loving family, CRN friends, family and medical experts looking out for her, Josie will continue to pursue those activities that bring her joy and will share her sunny disposition with those she meets. “I’m just enjoying being me,” she said at her home last week.

Jenni, Jim and daughters, Josie, (left) and Jordan Sexstone recently traveled to Washington D.C. during Rare Disease Week on Capitol Hill, February 28th to March 1st. They brought the needs of the rare disease community and the cystinosis community to their congressional representatives during Lobby Day discussions.

(EXAMINER PHOTO | MANDY LOEHR)
We recently celebrated Rare Disease Day, an event that aims to bring awareness to the existence of rare diseases, celebrate the progress that has been made in rare disease treatments, and highlight the continuous need for research and therapy development for those diseases.

Nowadays, this initiative has a global impact, with events happening in over 80 countries; but back when Marybeth Krummenacker’s daughter was diagnosed with cystinosis, the push for rare disease research and treatment was only beginning. Marybeth Krummenacker was nominated as a Rare Disease Difference Maker™ by Jean Campbell of JF Consultants LLC for her role in founding one of the earliest rare disease patient advocacy groups, the Cystinosis Research Network, and for fighting to get her daughter Laura proper treatment during a time when there was little knowledge surrounding the condition.

Marybeth first became involved in the rare disease community when her daughter was diagnosed with cystinosis over 29 years ago. Soon after Laura’s diagnosis, she started to meet other families who had children with the same condition. She recalls, “We instantly became bonded over our shared situation. The best part of it... was that someone finally understood.” The support that she and her daughter received stuck with her, and Marybeth says that she learned how important it was for both patients and families impacted by rare diseases not to feel alone.

After realizing the need for a support network for cystinosis patients, she also learned how to navigate all of the red tape to set up and administrate a non-profit organization. She learned from the AIDS and breast cancer movements, which at the time were just
starting to gain traction, that the squeaky wheel gets the grease. “You can’t be afraid to open your mouth, that’s the thing,” Marybeth implores, “You have to be your own best advocate. You just have to be empowered.”

She learned this lesson, too, through her daughter’s diagnosis. When she first noticed something was wrong with Laura, her pediatrician told her that she was just an over-reactive mother. “I was told she would outgrow it, that she was fine. I was dismissed.” Marybeth was angry. She knew in her gut there was something wrong, and she had to learn quickly to find her voice because no one else was going to do it for her. It was at a NORD meeting where she first met the doctor who would diagnose her daughter. After hearing of some of Laura’s symptoms, instead of dismissing her like other pediatricians, he said three words that changed the course of her and her daughter’s life: “Tell me more.”

In the rare disease community, often it is a tandem effort between the government, pharmaceutical industry, and families to push for the commercialization of treatments for rare diseases, and this situation was no different. The doctor, along with several other physicians, the NIH, and biopharmaceutical companies worked together to bring to fruition the FDA approval of several treatments for cystinosis in the years that followed. Marybeth has a close working relationship with her local elected officials, sat on the board of directors at NORD for 6 years, and credits the positive progress of cystinosis treatments to the introduction of the ODDA initiative. This familiarity with both the patients and the politicians is the reason behind her success as a patient advocate.

Times have changed quite a bit since Marybeth first became involved in the rare disease community. Her role in starting the Cystinosis Research Network in order to advocate for her daughter and others demonstrate exactly why she stands out in the rare disease community as a Rare Disease Difference Maker™. Although the rare disease community has made significant progress over the years in all aspects, the one thing that hasn’t changed is the driving force behind the patient advocacy organizations: the parents.
May 07, 2018

This year our community celebrated the first annual Cystinosis Awareness Day (CAD) on May 7th. May 7th (5.7) was chosen to represent the most common cystinosis mutation: 57-kb deletion. The goal was to educate our family, friends, healthcare teams and general population about cystinosis.

It is estimated that at least 2,000 individuals worldwide have cystinosis, though exact numbers are difficult to obtain because the disease is often undiagnosed and/or misdiagnosed. Now is the time to improve diagnoses, improve treatments, advocate for our children and ourselves, and become relentless in our mission until a cure becomes a reality.

Thank you to everyone who made this inaugural event a success! A great way to continue spreading awareness year-round is to tell your story. If you are comfortable, tell friends, family, classmates and colleagues what cystinosis is and how it has impacted your life.

Planning for the 2019 Awareness Day will begin soon. Join the task force by emailing Clinton at cmoore@cystinosis.org. Some ongoing activities to support awareness and research include:

Fundraising. Start your own cystinosis fundraiser or piggyback on an existing Cystinosis Awareness Day event. This can be a walk/run, gala, golf outing, or online donations via your Facebook page.

Donate. Help support finding a cure for cystinosis by making an online donation or mail a check payable to "Cystinosis Research Network" to Cystinosis Research Network, 302 Whytegate Court, Lake Forest, IL 60045 USA.
Cystinosis Network Europe (CNE) Hosts First Cystinosis Conference in Berlin

Christy Greeley, CRN Executive Director and Jen Wyman, CRN VP of Family Support, look forward to attending and participating in the upcoming CNE meeting in Berlin as representatives of CRN and the entire U.S. cystinosis community.

The Cystinosis Network Europe (CNE) is a network of Cystinosis Patient Organizations of Belgium, France, Germany, Ireland, Italy, Spain, the Netherlands and UK. As you might already know, this network was founded in 2016 as a partner in the European and worldwide network of patients, caregivers, healthcare providers, researchers and others.

In July 2018 CNE will organize for the first time a European Cystinosis conference in Berlin. You can find more information about the conference on our website following this link https://www.cystinosis-conference-berlin-2018.eu/.

The conference will be focused on adults’ living with cystinosis, focusing on their questions, problems and research, but will also cover topics like: eyes, bones and muscles, hormone therapy for men and women, neurological or orthopedic problems that are of interest for everybody in the cystinosis community. We will offer round table discussions for different age groups and topics. The presentations will be held by specialists accompanied by a patient or parent to offer a lay perspective to problems in the “real world”.

Europe is a large and multicultural continent with a lot of different languages. We are happy to offer simultaneous translation for German, French, Italian, Spanish and Dutch.

Of course, we are excited to see how many people are interested and registered already. 120 participants are registered (incl. 40 adult patients) from all over Europe and the

See you there!
United States. 25 international specialists are confirmed as speakers along with a large informative poster session just waiting for visitors. So, this is an excellent opportunity to hear all

the cystinosis research news and meet more patients in one place than a clinician and specialist ever thought possible to see and speak with. Patients will have the chance to meet and get to know others with cystinosis better and learn how others manage their lives. This is one of the most important and life altering parts in our conferences, the ability to connect with those who know what living with cystinosis means.

Registration is open, please visit our website https://www.cystinosis-conference-berlin-2018.eu/. The earlier you register, the easier it is for us to plan with conference rooms, child care, and attractions for adolescents. We are waiting for you and are very happy to welcome everybody in Berlin. For further information feel free to use the contact given here or on the website.

This conference is organized and paid without the help of any pharma company. We can say: Our CNE members raised enough money to meet all the costs. We are free in what we offer and say, not relying on refinancing.

Warm regards and thank you,
Claudia Sproedt M.A.
Cystinosis Network Europe

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Eloxx Pharmaceuticals Announces FDA Grant of Orphan-Drug Designation for Investigational Drug ELX-02 for Treatment of Cystinosis

April 27, 2018 12:41 ET | Source: Eloxx Pharmaceuticals

WALTHAM, Mass., April 27, 2018 (GLOBE NEWSWIRE) -- Eloxx Pharmaceuticals, Inc. (“Eloxx”), (NASDAQ: ELOX), a clinical-stage biopharmaceutical company dedicated to the discovery and development of novel therapeutics to treat cystic fibrosis, cystinosis and other diseases caused by nonsense mutations limiting production of functional proteins, today announced that it has received orphan-drug designation from the Food and Drug Administration (FDA) for ELX-02 for treatment of cystinosis from the FDA and that it will ring the Nasdaq Closing Bell on Monday, April 30, 2018.

“We are pleased to have received an orphan-drug designation for ELX-02 for treatment of cystinosis and for the opportunity to ring the NASDAQ Closing Bell in celebration of our recent listing on the Nasdaq coincident with our successful $50 million public offering,” said Robert E. Ward, Chairman and CEO of Eloxx Pharmaceuticals. “As we ring the closing bell, I would like to acknowledge and thank the employees, investors, patients, and partners who have supported Eloxx in our journey thus far. Given the clinical progress in 2017 for our lead product candidate, ELX-02, we are poised to seek regulatory clearance to initiate Phase 2 clinical trials in cystic fibrosis and cystinosis this year in Belgium and the United States, respectively. Our listing on the Nasdaq and capital raise position us well to advance these and other future clinical programs and pursue our mission, which is to transform the lives of patients with rare and ultra rare diseases.”

Eloxx’s common stock trades on the Nasdaq Global Market and began trading under the symbol “ELOX” on Thursday, April 26, 2018. A live webcast of the honorary opening bell ceremony, courtesy of Nasdaq, will begin at 3:45 am ET on Monday, April 30, 2018, and will be available at https://new.livestream.com/nasdaq/live and also on the Investor Relations portion of Eloxx’s website at www.eloxxpharma.com.
The 5th Annual Chandler’s Chance Fundraiser for CRN

By Clinton Moore

The 5th annual 2017 Chandler’s Chance Christmas fundraiser was an absolute success!! We had great weather, many activities, and most importantly, record breaking attendance numbers. This year we had just over 600 attendees which included 16 cystinosis families.

The night was filled with a variety of activities including fire truck rides, hay rides, Mr. And Mrs. Santa Clause, raffles, a 50/50 drawing, a live band, a food truck and so much more.

The exciting thing about a growing event is that you see new faces which means you are raising new awareness. Telling people about cystinosis that have never heard of it before. And then they go tell their friends and family and it just keeps growing.

A very special thank you to all the people who attended, donated, and volunteered their time and services to make this event so successful. Without all of you there would be no way to host an event of this size.

Attendees at the 5th annual Chandler’s Chance fundraiser supporting CRN. There were 16 cystinosis families in attendance this year.

This awareness ribbon, designed and built by the Moore family, represents all those living with cystinosis (green lights) and all those who have passed (white lights) worldwide. It was the inspiration behind CRN’s rare disease awareness ribbon this past February on Rare Disease Day.
National Pediatric Transplant Week - April 23-27, 2018

Donation and Transplantation Statistics

- Currently, nearly 2,000 children under the age of 18 are on the national transplant waiting list. More than 500 of the children waiting for transplant are between 1 and 5 years old.

- More than 1,800 children received transplants in 2017.

- There were nearly 900 pediatric organ donors in 2017.
- The donors ranged in age from newborn to age 17, most were between 11 and 17 years old.
- In 2017, more than 120 pediatric organ donors were babies under the age of 12 months.

- The size of the recipient's body and the donor organ are taken into account when matching donors to recipients. Very small children most often receive donations from other young people – although older children and adults can also be a good match.

- Sometimes, children can receive deceased or living donations of partial organs, like a portion of a liver.

- Most children under the age of 1 year are waiting for a heart or a liver.

- Most children age 1 to 5 years are waiting for a kidney, liver or heart.

- Most children age 5 to 10 years are waiting for a kidney.

- Most children age 11 to 17 years are primarily waiting for a kidney, followed by liver and heart.

- More than 138 million people, approximately 56% of the U.S. adult population, are registered organ, eye and tissue donors.*

- Pediatric donation: A parent or legal guardian must authorize the donation for anyone under the age of 18 years. 15-17 year olds may register their intent to be a donor; however, until they are 18 years old, a parent or legal guardian makes the final donation decision.

- To register your decision to save and heal lives, visit RegisterMe.org. To learn more about organ, eye, tissue and living donation, visit DonateLife.net.

* Living donation is not included in a donor registration.

Data from the Donate Life America Quarterly Donor Designation Report and the Organ Procurement and Transplantation Network (OPTN) as of March 19, 2018.
Sarah Larimore’s Transplant Journey

By Helen Adams | adamshel@musc.edu | December 19, 2017

Sarah Larimore, a 12-year-old girl with pink streaks in her blonde hair, smiles as a young woman is rolled into her hospital room at MUSC Children’s Health in a wheelchair.

“There’s somebody who wants to see you,” transplant surgeon Satish Nadig tells Sarah. That somebody is Heather Cox, who has never met Sarah before now — but whose decision to become a living donor and give a kidney to a stranger has changed Sarah’s life.

Sarah’s mother, Katie Larimore, rises to hug Cox. “I can’t even thank you enough. This is our Christmas miracle.”

Sarah was diagnosed with cystinosis as a toddler. It’s a recessive genetic disease. That means it only shows up if two people who carry the recessive gene for it have a child together. And even then, the odds are just one in four that the child will have cystinosis. Sarah was that unlucky one.

Cystinosis causes the amino acid cystine to build up in the body, often causing end-stage kidney failure before a child’s 10th birthday. Sarah’s kidneys were so badly damaged by scarring from the disease that both were removed in August. She’s been on dialysis since then, a process that does the blood-purifying work that kidneys normally do.

The Blythewood, South Carolina, girl ended up at MUSC Children’s Health, about two hours away from her home, because it has the largest kids’ kidney treatment program in the state and is ranked a “best hospital” for nephrology by U.S. News & World Report for 2017-2018.

Heather Cox was familiar with the challenges of kidney disease. Not her own, but her boyfriend’s. Ken MacGillivray, who has type 1 diabetes, had kidney and pancreas transplants at MUSC Health in July. Cox took care of him while he recovered.
She also made an important decision. Cox told the living transplant donor coordinator at MUSC Health, Kimberly Goad, that she wanted to give one of her kidneys to someone who needed it. She wanted that person to have the same fresh start as MacGillivray had. There was one condition: Cox wanted the organ to go to a child. Through the United Network for Organ Sharing, or UNOS, she was matched with Sarah.

Sarah and her mother weren’t far away. They’d rented a condo in Mount Pleasant to be near MUSC Children’s Health for treatments while they waited for a donor. Sarah’s mother remembers the day they learned about Cox’s generous offer. “It was amazing. I was driving across the Cooper River Bridge when Kimberly [Goad] called me, and I thought I was going to drive off the bridge. I was crying and couldn’t see where I was going,” Katie Larimore says.

The MUSC Children’s Health transplant team was thrilled to find a match for Sarah, too. Its program is the busiest in its UNOS region, but the team is not too busy to pause to recognize the special impact each transplant has on multiple lives.

So when the doctors, nurses and other transplant specialists found out Cox and Sarah were about to finally meet, they flocked to Sarah’s room to witness the meeting between a woman willing to literally give part of herself to a stranger and the sixth-grader whose life she helped save.

In Sarah’s hospital room, tears flow as Cox rises from her wheelchair with assistance. “I want to hug peanut,” she says, referring to Sarah. She carefully walks the few steps separating her from Sarah and leans down to embrace her.

“Thank you so much,” Sarah says. “We’re so happy for you baby,” Cox answers. “You’re going to do great.” Sarah’s parents are smiling as tears roll down their faces. Her mother tells Cox they don’t know what to say about what she’s done for Sarah. “Your kidney is beautiful,” Katie Larimore says. “Dr. Nadig is bragging about her labs. She’s perfect. She’s hurting, but it’s worth it.” Her husband, Jeff Larimore, agrees. “She’s thriving.”

Cox has already sent a stuffed bear to Sarah, which lies on Sarah’s bed. Sarah’s family gives Cox a quilt made for her by a family friend. “Thank you for my blanket,” Cox says. I love it.”
They also talk about something else Cox and Sarah have in common: brightly colored hair. "I can’t believe Heather has blue hair. It’s the coolest thing ever," Katie Larimore says. "Sarah has always had colored hair. It’s her thing."

Katie Larimore then describes what their family has been through. "We’ve been waiting for 10 and a half years," she says. "All kids with cystinosis need a transplant, and do very well."

She knows exactly what she’s talking about, thanks to the family’s involvement with the national Cystinosis Research Network. Both Katie and Jeff Larimore have served as volunteers, meeting other families and raising funds and awareness about the disease.

Due in part to that organization’s hard work, there’s good news for the estimated 2,000 children around the world with cystinosis: Treatment is more widely available than it used to be, and children with the disease can live into adulthood.

Sarah will still have to take medication for cystinosis, along with anti-rejection drugs to keep her body from rejecting the new kidney, but Cox’s donation means Sarah will be able to get back to enjoying all the things a 12-year-old girl should. And maybe more, after all she’s been through, her parents say.

"I want to swim with dolphins again. In the Bahamas," Sarah says.

Her mother answers, "You want to go? I guess we’re going."

Sarah follows up. "You said we could go on a cruise."

"We’ll do it," her mom agrees.

Sarah isn’t the only one with something to look forward to. The Larimores tell Cox that she needs to check out Facebook, where the cystinosis community has been following Sarah’s situation closely.

"We know everyone in the United States with this disease, but also have a huge international community," Katie Larimore says. "If you’re on Facebook, you can friend me and read all the well wishes you have received from around the world. A story like this, where one of our patients is having a miracle, literally, it raises hope for cystinosis patients around the globe. We’ve got hundreds of well wishes for you.

"You have brought hope to 2,000 people with this disease around the world, and their faith is restored. Some people have been on the transplant waiting list a really long time. It’s not just Sarah that you’ve helped," she adds. "It’s our entire community."

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Public Affairs and Media Relations
135 Cannon Street, Suite 403, MSC 189, Charleston, SC 29425
Cystinosis Ribbon Awareness Sticker Now Available

We've created a sticker to help family and friends communicate the importance of raising cystinosis awareness and education through any gesture - big or small. (See link below to get yours)

https://cystinosis.org/news/announcements/250-rdd2018

This, however, is no ordinary sticker. It was inspired by a structure standing 12 foot (3.66m) tall. It was thoughtfully created containing 2,000 green lights, one light for each person living with cystinosis. The outline of white lights represent those who have passed but remain part of who we are today. This symbolic gesture has been captured in a ribbon sticker ready to travel the globe spreading awareness, education, support and love for you.

Once your ribbon arrives, start sharing! Post it for the world to see. If you are on social media, tag your ribbon photo with #mycystinosis to educate others about cystinosis and what it means to you (the good, the bad, and sometimes the ugly). Your post could help another family or individual trying to manage daily life with a rare disease. Cystinosis warriors...thanks to you!

Strength: Lives Touched by Cystinosis

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 Amazon.
DUBLIN CYSTINOSIS WORKSHOP 2018

This is the fourth year that Cystinosis Ireland has hosted the Dublin Cystinosis Workshop (DCW) which brings together world-class international scientists from a wide variety of disciplines to network, share ideas, discuss scientific breakthroughs and to work together with the common aim of conquering this rare disease.

This year, Cystinosis Ireland was delighted to welcome 22 scientists from New Zealand, USA, England, Scotland, Belgium, Netherlands, Germany, Italy and of course from Ireland (North and South) to DCW 2018 which was held on April 7, 2018.

Research topics discussed at the workshop included new drug therapy formulations and delivery strategies including the use of contact lenses for more effective ocular cysteamine delivery; understanding the molecular biology and pathophysiology of the cystinosis disease and various genetic strategies aimed at developing a long-term cure as well as discussions around issues of infertility and treatments of the disease that go beyond cystine depletion.

The workshop was moderated by Dr. James Murray, Trinity College, Dublin and Dr. Patrick Harrison, University College, Cork.

A key objective of this year’s workshop was to highlight important research questions that are relevant to cystinosis patients and their families but that are not well addressed by researchers yet.

2018 is the 15th anniversary of Cystinosis Ireland’s establishment and of its commitment to supporting world-class research into better understanding, treating, and finding a cure for the immensely challenging rare genetic disease that is cystinosis.
Cystinosis Ireland continues to seek new research opportunities in areas of interest that will bring us closer to improving the treatment of cystinosis, enhancing the lives of those living with the disease and taking one step closer to finding a cure.

Cystinosis Ireland also plays an important role in advocating for and providing support to those living with cystinosis on the island of Ireland.

We’ve accomplished a lot in the past fifteen years and we will continue to do so until we find a cure. We value and appreciate everyone who helps us in achieving these goals.

Acknowledgements

Hosting the 4th Annual Dublin Cystinosis Workshop would not be possible without the very generous support from public donations received by Cystinosis Ireland.

We would also like to acknowledge additional conference support received from the Health Research Board (HRB) and Science Foundation Ireland (SFI).

Cystinosis Ireland would also like to thank the scientific committee for their hard work in organising this workshop – Dr James Murray, Trinity College, Dublin, Dr Achim Treumann, University of Newcastle and Ms Anne Marie O’Dowd, Director Cystinosis Ireland, with administrative assistance provided by Dr Ruth Davis and Ms Denise Dunne.

On behalf of Cystinosis Ireland, Ms Anne Marie O’Dowd joined with representatives of other patient organisations, healthcare professionals, families and individuals who help promote organ donation and transplantation at a special St Patrick’s Day reception hosted by President Michael D Higgins and his wife, Sabina Higgins, at the President’s home, Aras an Uachtarain, in Dublin.

Highlighting the wonderful generosity of organ donors and their families at a time of great personal tragedy the President noted “Organ donation is an act of incredible solidarity, and perhaps the greatest gift one human being can give to another.” He encouraged everyone to have a conversation with their next of kin and let them know about their wishes to be an organ donor.
There are currently approximately 550 people in Ireland awaiting life-saving heart, lung, liver, kidney and pancreas transplants, who along with their families and loved ones are hoping to benefit from the powerful gesture of organ donation.

All of those attending the reception were treated to an afternoon of music, song and dance, lovely refreshments. Cystinosis Ireland wish to thank President Higgins and his wife for their warm welcome and for taking the time to nurture a public debate on this vital topic.

Visit to Aras an Uachtarain to honor those involved in Organ donation

Figure 4. Ms Anne Marie O’Dowd of Cystinosis Ireland (second right) pictured with the President of Ireland and his wife (centre) and members of other patient organisations attending the St Patrick’s Day reception at Aras an Uachtarain honouring all of those involved in organ donation and transplantation.
Deanna Lynn Potts Scholarship from the Cystinosis Foundation

Deanna Lynn Potts was born with Cystinosis and lived to be 27 years old. Before she died, she discussed her wishes to start a scholarship fund for children with Cystinosis.

We know how devastating a chronic illness can have on a family emotionally, physically, socially, and financially. Children with Cystinosis are living longer, thanks to medical science and therefore, embarking on careers.

These careers require education. Education is expensive, yet something we do not want to deprive our children of in our world today. Due to the financially draining medical costs, it might prove difficult to send a child to college.

Through this fund, we hope to help some of those students.

**PURPOSE:** To provide supplemental financial assistance to an undergraduate student diagnosed with Cystinosis enrolled in an accredited collegiate or vocational program.

**SCHOLARSHIP AWARD:** A $1000 scholarship awarded annually. The award is contingent upon the winners acceptance to an accredited college, university, or vocational program and will be payable to the education institution to be applied to tuition, room and board.

**ELIGIBILITY:** Each candidate must be a current high school senior, who has Cystinosis and have a financial need.

**APPLICATION PROCEDURE:**
Documentation/verification of Cystinosis (e.g. letter from physician.)
An official copy of high school transcript.
Two letters of recommendation from current teachers/faculty members and/or counselors regarding applicant's scholastic aptitude and personal qualifications.
An essay of 500 words. We want to know a person who has played a vital role in student's life. How? Why? The essay should be typewritten and double spaced
A copy of the previous year's income tax return.

**JUDGING CRITERIA:** The essay will earn a possible 40 points and will be judged on the basis of rationale, grammar and comprehension. Transcripts and letters of recommendation will carry a possible additional 20 points. The Cystinosis Foundation Board will establish an independent judging panel to evaluate and rate the applicants. The decision of the judges is final. Finalists may be interviewed before selection is made.

**DEADLINE FOR APPLICATION:** Application and all accompanying documents must be received at the Cystinosis Foundation Office in a single, flat package by August 15th of each year. You can call the Cystinosis Foundation for an application 888-631-1588 or you may open it by Clicking on the link below, Print the Application and mail it to the Cystinosis Foundation.

**Remember, the deadline for receiving applications is August 15th of each year.**

[APPLICATION FOR SCHOLARSHIP](pdf)
Future by Design, Talking with Todd

By Todd Bradley

Future by Design is a group of adults with Cystinosis who have a lifetime of experience and want to build better futures for everyone experiencing Cystinosis. The goal of our group is to bring voice to issues, develop initiatives, and build a strong, positive community. Since Cystinosis is not common and those of us who share the diagnosis often live far apart, our hope is that we can bring our community together and support people who struggle through the daily challenges of living with this disease. We have many initiatives that you will learn about in the coming months. First, we would like to introduce Todd Bradley, one the members of the Future by Design board. He has a video series that answers questions from the community and provides hope to our community.

Why did you want to join Future by Design (FbD)?

That is a great question.

I guess I wanted to join FbD because all my life I felt I had no purpose, no place to call my own. Future by Design is a group of people who really know the struggle of this disease called cystinosis. For me, this was an opportunity to leave my mark and lend my voice to others, so they can prosper and realize cystinosis isn’t something that has to dominate your life.

What does Future by Design mean to you?

Wow… one word comes to mind, FAMILY! I wish FbD was a part of my life decades ago. Life would have been so much easier in the past. I don’t even really count just my life, it would have been easier for so many others. Just imagine how many lives could have been impacted if FbD was in existence 10 years ago! I truly believe in my heart one day our cause will not only affect the cystinosis community in a profound way, but so many others who don’t have this disease. I know I would like to reach out to others who struggle from any debilitating disease and even someone who just needs to talk about their own personal demons. In all honesty, many of us in the world see vulnerability as a weakness. This is not a good way to live, and if we cannot openly talk about our own issues with each other, there is no way to move forward. We all deserve to be content and happy!

How can people utilize Future by Design?

Reach out! This is the most important first step! There are so many of us in FbD willing to talk and give advice. I understand life gets hectic and busy, but that is not a valid excuse. Do not be afraid, embarrassed, or uncomfortable with what you need to talk about. No question is stupid! We are all here for you, and they must take that leap of faith! We don’t judge or criticize; our group is here for everyone. The last thing I can say is, we all have our struggles with this disease, but if we do not speak out and seek help, things will get worse. Communication is key!
What can Future by Design do for others?

Well let’s see…. Let me first discuss what FbD did for me.

When I first joined FbD I was apprehensive, I thought it was useless. I asked myself “what in the world could I offer?” Although it involved other adults, I found out quickly that we all had this disease but our lives were very different. Some of us had it easier and some of us had a rougher time during our lives. The main thing I remember is that we all could help each other in so many unique ways, the support and advice was priceless. The friendships I made were going to last forever. I felt that for the first time my voice was valued. I must say I couldn’t believe that I was making not only a difference but I was also helping my community in a tremendous way. The uncomfortable subjects were no longer taboo, and everyone around me understood!

So, in conclusion, what can Future Bb Design do for others? FbD can do many things, but from experience it will help calm the storm in our minds, relieve the stress in our souls, and when someone goes to war with this disease, they need to know they have an unstoppable army behind them ready to give their hearts to help in any way!

Why did you choose to make the Talking with Todd video series?

That is the easiest question to answer! I make the videos because so many who struggle with cystinosis either don’t have a voice or choose not to speak up, and that is okay! My community is my life, and I will not shy away from any subject. Although I have lived with this disease for over 32 years it no longer scares me. I take any struggle full force! I will not let others face this alone. I have talked about some scary issues and I will continue to get deeper in it. My warriors have done so much for me to move forward and I will pay it back. We all deserve happiness! I will not let anyone face this struggle alone and I’m always willing to be a voice for any of you!

What do you hope to accomplish through the videos?

I know this isn’t a perfect world but if it was I would like to raise more awareness and to have everyone come together more than ever. When I meet or talk to others in the struggle I see the pain in their eyes and sometimes I even feel it. I see the little ones and they are so happy, but I see the parents and see the fear in their eyes! They are so overwhelmed! What I want more than ever is a cure! We must come together for that to come true! Life is tough for everyone no matter the disease or the ailment. I want everyone to realize we can’t ignore the reality and for research to continue we need to be the community we should be. Through my videos I hope to inspire and if I help only one single person I know I made my mark in this world. Our purpose as humans on this rock we call Earth, is to make it better for the next generation. I beg all of you to find your purpose and be happy, but most importantly help the community to better the lives that are here and the lives to come. I need you and we need each other.

Final Thoughts from Talking with Todd

It does not matter what Foundation or Network you follow or support. What does matter is the love you show in your heart. Life is precious and can be wonderful. Spread the word about cystinosis and other rare diseases! It is so important for all of us to feel that we belong and are loved. When things go wrong in our lives is it good to know that others have been there. No one can do it alone! Give yourself a chance to live in the most meaningful way. Stay strong my warriors, life can be so good when you believe in yourself and each other! Life is what you make it. It is up to you.

For Todd’s videos plus more check out the link below.
https://www.youtube.com/channel/UC3BCCVrDmY6M7ZKfUeBmOYQ/videos

If you are interested in joining the board and helping more people in our community, please contact Emily Mello, Mmhmm03@hotmail.com. Or, if you are looking for support or just want to talk to someone who has been through what you may be struggling with, take a look at our Outreach Program cystinosis.org/family-support/future-by-design.
Horizon Pharma is pleased to invite you to a program designed specifically for patients and caregivers living with cystinosis. This program is an opportunity for you to learn about living with cystinosis while connecting with others impacted by the condition. This dynamic, 4-hour, interactive educational program will include opportunities for you to:

- Understand the importance of continuous cystine control
- Learn about PROCYSBI® (cysteamine bitartrate) delayed-release capsules for the treatment of cystinosis
- Participate in discussions and exercises, including an informative presentation about PROCYSBI

Tentative meeting locations for 2018 are Columbus, Ohio; Detroit, Michigan; Seattle, Washington; Phoenix, Arizona; St. Louis, Missouri; Hershey, Pennsylvania; Atlanta, Georgia; New York, New York; Nashville, Tennessee; and Birmingham, Alabama.

To RSVP to one of these cities or to request a program near you, please contact Jennifer Caughlin at 602-953-2552.

Horizon Pharma is the sole creator and sponsor of IMPACT programs.

IMPORTANT SAFETY INFORMATION

What is PROCYSBI® (Pro-CIS-bee)?
PROCYSBI® (cysteamine bitartrate) delayed-release capsules is a prescription medicine used to treat a medical condition called nephropathic cystinosis, in adults and children 1 year of age and older.
It is not known if PROCYSBI® is safe and effective in children under 1 year of age.

Who should not take PROCYSBI®?
Do not take PROCYSBI® if you are allergic to penicillamine or cysteamine.

Please see additional Important Safety Information about PROCYSBI® on the following page.
IMPORTANT SAFETY INFORMATION (continued)

What should I tell my doctor before taking PROCYSBI?

Before you take PROCYSBI, tell your doctor if you:

- drink alcohol
- have a skin rash or bone problems
- have or have had stomach or bowel (intestinal) problems including ulcers or bleeding
- have a history of seizures, lack of energy, unusual sleepiness, depression or changes in your ability to think clearly
- have liver or blood problems
- have any other medical conditions
- are pregnant or plan to become pregnant. It is not known if PROCYSBI will harm your unborn baby. Tell your doctor right away if you think that you are pregnant. Talk with your doctor about the benefits and risks of taking PROCYSBI during pregnancy.
- are breastfeeding or plan to breastfeed. You should not breastfeed during treatment with PROCYSBI. Talk with your doctor about the best way to feed your baby if you take PROCYSBI.

Tell your doctor about all medicines that you take, including prescription and over-the-counter medicines, vitamins, and herbal supplements. Know the medicines you take. Keep a list of them to show your doctor and pharmacist when you get a new medicine.

What should I avoid while taking PROCYSBI?

Do not drive or operate machinery until you know how PROCYSBI affects you. PROCYSBI can make you sleepy or less alert than normal. Do not drink alcohol if you take PROCYSBI. Drinking alcohol while taking PROCYSBI may change how PROCYSBI works and may cause an increase in the amount of PROCYSBI in your blood that may cause serious side effects.

What are the possible side effects of PROCYSBI? PROCYSBI can cause serious side effects, including:

- Skin, bone, and Joint problems. People treated with high doses of cysteamine bitartrate may develop abnormal changes of their skin and bones. These changes may include stretch marks, bone injuries (such as fractures), bone deformities, and joint problems. Check your skin while taking PROCYSBI. Tell your doctor if you notice any skin changes or problems with your bones or joints. Your doctor will check you for these problems.
- Skin rash. Skin rash is common with cysteamine bitartrate and may sometimes be severe. Tell your doctor right away if you get a skin rash. Your dose of PROCYSBI may need to be decreased until the rash goes away. If the rash is severe, your doctor may tell you to stop taking PROCYSBI.
- Stomach and bowel (intestinal) problems. Some people who take other medicines that contain cysteamine bitartrate develop ulcers and bleeding in their stomach or bowel. Tell your doctor right away if you get stomach-area pain, nausea, vomiting, loss of appetite, or vomit blood.
- Central nervous system symptoms. Some people who take other medicines that contain cysteamine bitartrate develop seizures, depression, and become very sleepy. The medicine may affect how your brain is working (encephalopathy). Tell your doctor right away if you develop any of these symptoms.
- Low white blood cell count and certain abnormal liver function blood tests. Your doctor should check you for these problems.
- Benign intracranial hypertension (pseudotumor cerebri) has happened in some people who take immediate-release cysteamine bitartrate. This is a condition where there is high pressure in the fluid around the brain. Your doctor should do eye examinations to find and treat this problem early.

Tell your doctor right away if you develop any of the following symptoms while taking PROCYSBI: headache, buzzing or "whooshing" sound in the ear, dizziness, nausea, double vision, blurry vision, loss of vision, pain behind the eye or pain with eye movement.

The most common side effects with PROCYSBI include:

- nausea, stomach (abdominal) pain, breath odor, diarrhea, skin odor, tiredness, skin rash, headache, and problems with body salts or electrolytes.

Tell your doctor if you have any side effect that bothers you or that does not go away. These are not all of the possible side effects of PROCYSBI. Call your doctor for medical advice about side effects.

You may report side effects to the FDA at 1-800-FDA-1088.

For more Information about PROCYSBI, visit PROCYSBI.com.
Summer Slam Baseball Tournament Supports CRN with Fundraising Effort

By Zachary Fell

Why we chose to take over Summer Slam Baseball Tournament?

Baseball is a huge passion of mine and having enjoyed my experience playing baseball for Lincolnshire from the ages of 8 – 12, I saw an opportunity to give back to the baseball program. My dad started the Summer Slam Baseball Tournament “Summer Slam” in 2014 while he was still the Director of Lincolnshire Travel Baseball. Once he stepped away from the Director role, no one else was jumping at the opportunity to carry on the tradition. Therefore, it presented an opportunity for me to get involved and give back while gaining experience in leading and project managing through the organization of a community event. One of the biggest changes for this year’s tournament is the addition of the philanthropic component to the fundraising efforts. Previously the tournaments resulted in donations to maintain the North Park baseball fields and help offset the Lincolnshire Travel Baseball fees. While some of the fundraising efforts will continue to be donated to the baseball program, my mom challenged us to choose a philanthropic organization this year to donate a majority of the money to.

How did we chose Cystinosis Research Network?

When determining which philanthropic organization we wanted to support this year, there was one thing that we knew and that was that we wanted it to be a cause that was close to the community of Lincolnshire. Ultimately, the committee chose Cystinosis Research Network (CRN). We chose CRN as we know Jack Greeley as a Lincolnshire resident, Stevenson High School Senior and a future Marquette Golden Eagle. Additionally, Jack grew up playing Lincolnshire baseball within the community like myself and the committee members. We all voted to support him and others in their fight against the rare disease of cystinosis.

Fundraising/Sponsorship

In years past, the fundraising was done through field sponsorship, windflags and concession sales. This year we wanted to do more. We decided to add some additional sponsorship opportunities. These additional sponsorships included sponsorship of the entire park, a company logo on the game day baseballs, shuttle buses used between parking lots, homerun fences, scoreboards as well as online auction items. We are proud to say that many Lincolnshire business owners were more than willing to
contribute to our efforts. Some of our 2018 high-level sponsors include Horizon Pharma, Slammers Illinois Baseball & Softball, True Partners Consulting, AFN, and Christopher B. Burke Engineering Ltd.

Innovative Sponsorship

In our initial conversations with Jack Greeley, we discussed avenues outside of tournament fees, sponsorships and concessions that would be effective in spreading awareness of cystinosis. As a committee, we decided to have bracelets made to sell prior to and during the Summer Slam Tournament. Kevin Weasler, owner of Culvers has been selling them at both his Lincolnshire and Buffalo Grove stores. Additionally, Lincolnshire resident and personal trainer Scott Zilligen of ZFit also kindly offered to sell them at his fitness center on our behalf.

Day Fundraising @ Culver’s

To show additional support, the newly opened Culver’s in Lincolnshire hosted a day of Fundraising for Cystinosis. On Sunday May 6th, Culver’s donated 25% of their sales between the hours of 11am and 8pm to Summer Slam / (CRN). Our Summer Slam committee members as well as some additional volunteers were at Culver’s promoting the event throughout the day. Thank you to all who made the day successful.
Family Support Update

By Jen Wyman, Vice President of Family Support

"Let us put our minds together and see what life we can make for our children."
-Sitting Bull

Life is full of fortunate and unfortunate circumstances and it is a journey of unknown terrain. This is especially true when a family faces a rare disease diagnosis. Darkness can sometimes overshadow the light...but the light eventually comes through in the form of HOPE.

We are fortunate in 2018 to have so many avenues to connect with each other. We never have to suffer alone. We have a family support network that makes cystinosis not seem so rare. We have each other.

It is my pleasure to be back on the executive board of CRN as the VP of Family Support. Twelve years ago our daughter was diagnosed with cystinosis. Twelve years ago we gained a secondary family within the Cystinosis Research Network. The members of this group have become our lifelong friends and the few people in the world who will ever know exactly what we have gone through. CRN is the light for all of us and it delivers the HOPE we all need to press on.

Contact Jen Wyman at jwyman@cystinosis.org

The Wyman family, Matt, Jack, Kacy, Jen and Tim.
Order, Store, & Use

To order, please call:
Walgreens Specialty Pharmacy
1-877-534-9627
Mon–Fri, 8AM–8PM EST

Store unopened bottles in the freezer

Every 7 days remove 1 bottle from freezer and thaw for 24 hours before use*

*ONCE THAWED, NO REFRIGERATION IS NECESSARY AND DO NOT REFREEZE

Discard bottle after 7 days

You are encouraged to report negative side effects of prescription drugs to the FDA.
Visit www.fda.gov/medwatch or call 1-800-FDA-1088
Jack Greeley 18th Birthday Letter Fundraiser for CRN

By Christy Greeley

The Greeley Family once again celebrated Jack’s St. Patrick’s Day birthday sending out the 16th edition of his birthday fundraising letter. As is his way, Jack has faced many challenges in the past year with his characteristic grace, courage and positive attitude. His 18th year was especially challenging as he underwent an extensive orthopedic surgery this summer which has taken months of recovery. On the positive side, he has enjoyed a wonderful Senior year of high school and looks forward to attending Marquette University in the Fall. We cannot thank our friends and family enough for all of the support we have received, this year and since his initial diagnosis in 2001. Since 2003 we have raised over $350,000 in honor of Jack to support CRN and the cystinosis community, thanks so much to everyone who has taken part over the years.

Following is an excerpt from this year’s letter:

"Assume the position” might be some of the scariest words a parent could hear about a child. Imagine it for one of your kids; under the threat of police arrest, sirens glaring, legs spread, and hands likely cuffed...probably hopeless in the moment. Those of you who know our family’s plight might think we have had our hopeless moments with our son, Jack. Fortunately, that has never proven to be the case.

As most of you know, Jack has cystinosis, a rare metabolic, genetic disease afflicting more than 2,000 people worldwide. We run the Cystinosis Research Network (CRN) from our home. This disease has the potential to impact all of the organ systems in the body leading to kidney failure, muscle wasting, diabetes, blindness, pulmonary deficiency, hypothyroidism, and neurological damage. Jack has developed unusual and relatively rare orthopedic complications, but has been blessed with better than average kidney function. Albeit not the hood of a police car, the reality is that Jack has been placed in many compromised positions in his brief life as he approaches his 18th birthday this St. Patrick’s Day.

Despite these physical restrictions, Jack is too tough, too strong, and too resilient to be limited. In fact, he is the ultimate contortionist and might be one of the most flexible people we know, because he has no choice. Do not let his inability to place his hands flat on the ground from a standing position fool you. Life has forced him into many positions, both literally and figuratively. What are those positions?

When Jack was diagnosed around his first birthday in spring 2001, he became positioned for a lifetime of picking, poking, prodding, nagging, cutting, examining, you name it. A
chronic disease forces you to have discipline and vigilance tied to medical routines that were previously taken for granted. For Jack, this has added up to over 160,000 pills swallowed divided up every six hours for his main medication, Cystagon to take effect. This is roughly 25,000 medication intervals (including getting up in the middle of the night over 6,200 consecutive days). There have been around 55,000 eye drop applications and well over 500 medical appointments. Jack has also faced surgery five times, flat on his back, knocked out, so that doctors could address ongoing issues: once to have a gastric feeding tube inserted into his stomach because he did not eat solid food for 18 months; twice for knee and ankle surgeries to help straighten these body parts where staples and screws were inserted; and twice for highly invasive orthopedic surgeries to help straighten his lower left leg.

Lots of literal medical positioning took place with these last two surgeries this past summer. Three procedures took place at once where six overall incisions and cuts were made to move and align his knee, leg, heel, ankle, and foot:

- **Triple C procedure** – three bones in his left foot were surgically cut and shifted with cadaveric wedges inserted to straighten the foot.
- **De-rotation Osteotomy of the tibia** – the tibia and fibula bones were cut and rotated to correct alignment and then secured with a metal plate and screws.
- **Installation of an eight plate** – his left knee (for the third time) had another eight plate inserted to take advantage of any potential growth remaining to further straighten the leg.

After exhausting the utility and usefulness of braces, physical therapy, and lesser surgeries over the years, we had to take these steps and undergo full reconstruction of Jack’s foot and ankle. Through growth hormone, testosterone therapy, and mega doses of phosphates and calcium to help strengthen Jack’s bones, he also grew a foot in the past three years. Now at his 18th birthday, Jack has been in many and will continue to be in a variety of fascinating positions:

- In the aftermath of these most recent surgeries, Jack was non-weight bearing for a month and limited weight bearing for another month. Imagine a 6-1, 200 lb. kid with pre-existing mobility limitations now faced with life in a La-Z-Boy. How do you sleep? How do you get up? How do you go to the bathroom? How do you shower? How do you eat? How does anyone in the house get anything done other than to help Jack? There were many funny positions for all. The good news? You can play A LOT OF VIDEO GAMES from a La-Z-Boy recliner!

- There needs to be some color commentary on Jack’s recent stint in a La-Z-Boy. Jack has received the gift of humility and simplicity. This has served him well his entire life and at no time was it more noticeable than this past July and August in recovery. Remember, Jack has an underlying kidney issue and drinks a lot of water. This put him in an, um, ah, shall we say compromised position over and over again. He became very adept at using a walker to pop up from his recliner, balance on his right leg, raise his left, braced leg, slide down his shorts, and pee into a hospital-issued urinal. But think about it; two hands on the walker, balance on the right leg, and elevation of his left leg. Who is holding the urinal? Ha! It is called a “total team effort” with assists from Mom, Dad, and then 20-year-old sister, Alex.

- We have learned one basic axiom applies to life – you just never know. For years, given Jack’s unusually challenging orthopedic issues, we theorized that it is possible some additional underlying problem is in

Jack Greeley celebrates his 18th birthday.
play beyond cystinosis. Jack has been tested and seen doctors coast-to-coast. Recently, the National Institutes of Health in Washington, DC followed up with us after some previous testing. They have reason to believe that Jack is in a very tiny subset of patients who do not have the gene deletion seen in virtually all cystinosis diagnoses. The conclusion – we know he has cystinosis, but we do not know why. So it looks like we will be finding ourselves in a new position by getting to the NIH in the coming months to begin some experimentation with their acclaimed Undiagnosed Disease Network Program (see the attached feature from “60 Minutes,” https://www.youtube.com/watch?v=Trb9mPcpVv4). Knowledge is power, so we look forward to it!

- This fall, Jack will be a college freshman at Marquette University in Milwaukee. While we welcome the opportunity, this is a major new change for all of us. How will Jack do on his own? How will he adhere to his 24/7 medical regimen? Will he take care of himself? We welcome this new position for everyone. Yes, it is a good school close to home, but Jack might be the most excited because they do not have a football team, the school’s initials contain a “U” and an “M,” and their colors are pretty much maize and blue. Easy transition!

Jack has faced many challenging positions and he most assuredly will have others as his adult life unfolds. He offered us a simple reminder during his surgical recovery why he will be just fine. After Jack got out of his La-Z-Boy, he began to sleep in his bed. While making it one day, we realized something pretty unusual. All night, Jack would sleep flat on his back with his left leg in a brace and elevated by pillows. He never moved during the night...NEVER...he could not do it. We do not know about you, but we would be miserable – stuck in one spot all night, no movement, no pillow fluffing, just pure immobility. With Jack, there was never one complaint. Not with the surgery. Not with the La-Z-Boy. Not with the statuesque sleeping position in the bed. Nothing. Why? Because the kid never complains about his position in life. NOT EVER. What a gift...what an inspiration...and that’s why he is The Champ and we celebrate his birthday each year with this letter.

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:
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2. Select Your Account
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4. 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.
By José T. Morales

The Cystinosis Research Network recently participated in The Life Sciences Patient Congress, held in Philadelphia, Pennsylvania. The Congress was focused on breaking down silos to incorporate patient voices and to drive innovative solutions, form essential stakeholder partnerships and enhance patient health outcomes. Participation came in the form of Clint Moore’s delivery of the Opening Patient Address and CRN representative José Morales, along with Clint, engaging in the four principle specialty tracks.

**Patient Adherence and Engagement Summit** – providing insights on how to develop targeted adherence and engagement strategies to motivate patients, improve health outcomes, and encourage stakeholder collaboration

**Patient Centered Clinical Trials Summit** – insights on how to build partnerships with patients to optimize the research and development process, clinical trial design and improve speed to market

**Patient Advocacy Summit** – providing guidance on how to put patients at the forefront of advocacy by fostering stronger relationships and transparent communication

**Patient Journey Mapping For Speed To Therapy Summit** – defining techniques to map the patient journey, shorten time to fill, and overcome barriers to patient access

There is a movement in the pharmaceutical industry to prioritize value-based care with a focus on patient outcomes and quality. The challenge it faces is how to build innovative solutions with transparent stakeholder engagement across a patient’s entire journey. Clearly, these are lofty goals and not achievable without considerable resources and effort.

The patient congress brought over 250+ industry leaders from life science, pharmacy, health plans, provider organizations, patient groups, and patient leaders to share innovative ideas and strategies, drive innovation, and create a community of stakeholders united to provide value-based care to patients.

CRN remains committed to representing our community with a strong voice and advocating for advancements across the entire spectrum of providers.
Research Update

By Christy Greeley, Vice President of Research

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 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.

Our Medical Advisory Committee has been hard at work updating CRN’s educational materials. Look for them to be available in the coming months on the website and at educational events.

Cystinosis at the National Institutes of Health

As a reminder, patients may contact the National Institutes of Health to be enrolled in the cystinosis protocol and for consultative care by Dr. Bill Gahl and Dr. Galina Nesterova. For more information, please contact:

Joy Bryant
(301) 443-8690
bryantjo@mail.cc.nih.gov

or

Galina Nesterova, MD
nesterovag@mail.nih.gov
Project Updates

**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. Following is a brief update on their project:

The aim of our research study is to understand how changes at the level of cells in the brain due to lysosomal storage dysfunction translate to changes in cognitive function and behavior. To study the neurobiological underpinnings of cystinosis, the lab uses a Ctns -/- mouse line which has a cystinosis, nephropathic, targeted mutation 1 in the gene. To do this, we have developed new technologies from recording the electrical activity of the brain in this model. Specifically, we have deployed a method for studying the physiology of individual neurons throughout the brain in awake behaving animals. This cutting edge research approach will allow us to identify the neurophysiological changes that arise from the Ctns -/- mouse and relate this to alterations in global brain dynamics. The ultimate goal being to discovery biomarkers of the disease and identify potential targets for subsequent intervention. In the first 6 months of this research project, we have successfully recruited and trained an MD/PhD student with interests in Neurology and development to perform surgeries and record from control mice to characterize baseline patterns of activity throughout the brain for comparison with the Ctns -/- mouse.
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 Levtschenko (Principal Investigator)
Katholieke Universiteit Leuven, Belgium

Year Two: $88,493
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.

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
At the Cognitive Neurophysiology Laboratory we are looking at perceptual and cognitive function in cystinosis using cutting-edge non-invasive electrical brain imaging (scalp recorded electroencephalograms EEG) and behavioral measures. Over the past year we made a major push on patient data collection. Drs Foxe and Molholm attended the Cystinosis Family meeting in Utah with post-doctoral fellows Drs Anna Francisco and Katie Behar, and research technician Douwe Horsthuis. Over the course of two days, the team participated in the meeting, met families, and performed EEG and cognitive testing in 5 children with cystinosis. In addition, over the past year we enjoyed hosting 25 families from around the country, who came to participate in research at our lab in the Bronx, at the Albert Einstein College of Medicine. Many took advantage of this opportunity and took an extra day to do some site seeing and shopping in Manhattan! Both children and adult patients have participated in our studies-- This is great because we can now use the brain measures to look at how perceptual and cognitive functions change over development in cystinosis. Shout out: We need a few more adults to participate before we can draw any conclusions!

We have a lot of data that we are processing and making sense of. So far, the cognitive and brain data are telling an interesting story:

**Multisensory Integration:** Our brains receive information from the environment through our senses including eyes, ears, nose and skin. Indeed, it is well known that our brains contain specialized regions that deal with the information received from our individual senses. However, research over the past decades has also shown that our brains put together or integrate the information from our different senses. This is called multisensory integration. For example, when we try to understand someone talking in a noisy environment it helps tremendously when we are able to see the speakers face movements. Multisensory integration works really well in cystinosis. This is good news, and contrasts with the finding that multisensory integration is impaired in a number of other clinical groups, including Neiman Pick C Disease and schizophrenia.

**Visual Processing:** We find that basic sensory processing differs in this group. Our brain data show that the visual response is substantially larger in individuals with cystinosis, and this is the case at all ages that we have tested. This presents a powerful biomarker that can be used to see if a treatment normalizes brain function. This may reflect reduced neural inhibition in visual cortex, and may be related to impaired visuo-spatial ability that has been observed behaviorally in cystinosis. Analysis of data from another one of the studies we collected data for will help us to test this explanation. Better understanding of the basis of this difference will provide clues for treatments that will normalize brain function.

**Executive Function:** We are also studying executive function, which is often affected in cystinosis. One assay looks at the ability to withhold a prepotent response. Here we see that there is a lot that works really well in cystinosis, in children and adults alike. Individuals with cystinosis can withhold their responses just as well as healthy controls. However, they make a few more errors, withholding responses when they don’t need to. When we look at their brain activity, we see that when they withhold a response, this effortful process engages the brain for a much longer period than in controls. Cognitive tests of executive function shows that these individuals can perform the tasks just fine, but that it takes them a little bit longer. We are performing additional analyses of the brain data to fully understand the implications of the prolonged processing on effortful trials.

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.
Clinical Trial for Distal Myopathy in Nephropathic Cystinosis Announced

Hello,

I am writing to tell you about a multi-part research study I am conducting at Massachusetts General Hospital, titled Clinical Trial Readiness for Distal Myopathy in Nephropathic Cystinosis. The purpose of this portion of the study is to create a disease-specific assessment tool for the cystinosis patient community called Distal Myopathy Function Score (DMFS). Your input is valuable and will eventually validate this cystinosis tool. Developing the DMFS is a step-wise process, and we may send an updated DMFS survey as we continue to refine our tool in the near future.

I am informing you about the opportunity to participate in this study because you are engaged in the cystinosis community. The Cystinosis Research Foundation is helping us recruit patients and distribute information about the study through social media and other platforms.

If you choose to participate in the study, please email the research team at rithuong@partners.org. We ask that you provide your name, date of birth, and email address. This information will be kept on a password-protected computer in a locked office at Massachusetts General Hospital for participant contact documentation. By doing this, we will do our best to protect your confidential information. Still, there is always a small risk of an unintended breach of confidentiality.

You will receive an emailed link to the survey. The DMFS survey should take you about 3-5 minutes to complete. We estimate that we will distribute this survey to 50-100 patients. Your answers are anonymously recorded in our system. The survey that we are sending you is specific to your email address and allows us to verify whether you have completed the survey. We may ask that you complete the survey additional times as we revise the DMFS based on periodic data analyses.

You will not receive any personal health benefits as a result of your participation in this research study. We hope that the results will benefit the treatment and care of patients with cystinosis in the future. Your participation is completely voluntary. Whether you participate or not will have no effect on the medical care you receive here at
Massachusetts General Hospital or Partners Healthcare, either now or in the future, and will not impact any benefits you receive now or have a right to receive. Your survey will not become a part of your medical record, and your completed anonymous survey will only be accessible by the study staff.

Please contact the study coordinator Rachel Duong at (617) 724-1330 if you would like to learn more about the study.

If you’d like to speak to someone not involved in this research about your rights as a research subject, or any concerns or complaints you may have about the research, contact the Partners Human Research Committee at (857) 282-1900.

We are required by the Health Insurance Portability and Accountability Act (HIPAA) to protect the privacy of health information obtained for research. This is an abbreviated notice, and does not describe all details of this requirement. During this study, identifiable information about you or your health will be collected and shared with the researchers conducting the research. In general, under federal law, identifiable health information is private. However, there are exceptions to this rule. In some cases, others may see your identifiable health information for purposes of research oversight, quality control, public health and safety, or law enforcement. We share your health information only when we must, and we ask anyone who receives it from us to protect your privacy.

Please contact my research coordinator Rachel Duong at (617) 724-1330 with any questions about this research study.

Sincerely,

Grace

Florian Eichler, MD
By Terri Schleuder

It was another very successful PAS exhibiting experience for CRN in beautiful, friendly downtown Toronto. This huge city is a juxtaposition of old and new, Victorian homes next to modern skyscrapers. The perfect location for the 2018 Pediatric Academy of Pediatrics meeting.

We welcomed many physicians and other attendees to booth 323 where we handed out educational information both in hard copy and on our flash drives and shared our cystinosis stories.

It was especially gratifying to be here during this week among 1,000s of pediatricians as we celebrated Cystinosis Awareness Day on May 7th, 2018. Many helped raise cystinosis awareness by their willingness to be photographed holding our cystinosis awareness ribbons and fliers.

Thank you!!

A wonderful surprise occurred when a member of our cystinosis community arrived at our booth. Kole Binger, recently graduated from college, is headed to medical school. She was a recent recipient of the Sierra Woodward Sibling academic scholarship and is a very impressive young woman, whose life and future has certainly been molded by her personal experience with cystinosis.
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.

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.

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
CRN is off to a great start in 2018. We sent eight individuals/families, sixteen in all, to Rare Disease Week on Capitol Hill from Feb. 25th to Mar. 1st. Among the many activities was the opportunity to lobby congressional members about the needs of the rare disease community by sharing their cystinosis stories. (Read more about RDW on pages 6-8.)

Out of that experience, democratic senator Chris Coons from Delaware, offered to sponsor a national resolution declaring 5/7 to be the first Cystinosis Awareness Day. This date was chosen because the most common cystinosis mutation is the kb 57 deletion. We are working hard to get a republican senator to co-sponsor this resolution as well.

In the meantime, CRN president, Clinton Moore, walked 57 consecutive miles across in his state of Delaware to celebrate this day. The purpose was to raise funds and bring awareness to cystinosis nationally. (Read more about Clinton’s walk on pages 10-13.)

CRN has announced the 2019 Family Conference will be in Philadelphia at the Philadelphia Hilton at Penn’s Landing from July 18-20. Plan now to attend and look for more details over the next several months on our website.

Please visit CRN’s updated website and see our new logo. Thanks to Christy Greeley and Clair Johnstone for their many hours of effort into this rebranding project for CRN. Our brochures and exhibit display will be updated as well reflecting our new look.

In May, CRN exhibited at the Pediatric Academic Society meeting (PAS) held in Toronto, Canada this year from May 5-7. Booth 323 was well represented by Carol Hughes, and Terri Schleuder. In addition to handing out the usual materials we did our best to support Cystinosis Awareness Day by handing out fliers and awareness ribbons to attendees. (Read more about the PAS on page 54.)
CRN has announced the deadline to receive 2018 Scholarship applications is August 15th, 2018. Completed materials need to be mailed and received by that date to be considered. Please mail to:

Terri Schleuder  
40472 Franklin Mill St.  
Novi, MI 48375

Scholarship applications and information can be accessed on CRN’s website at: https://cystinosis.org/family-support/scholarships

Please check out page 35 with an additional academic scholarship opportunity offered in Memory of Deanna Lynn Potts through the Cystinosis Foundation.

We’ve have many fundraisers and family stories scattered throughout the newsletter. Please be sure to read about them.

On a personal note I would like to mention that my term as the VP of Education & Awareness on the executive committee will end in July. It has been a great honor serving as a CRN board member over the last seven years in the roles of Secretary and the VP of Education & Awareness. I will be stepping down at the end of my term to pursue other interests and obligations. New energy and ideas are always valuable to an organization and I know whoever steps into this role will bring both. I look forward to remaining active in the cystinosis community in the years to come. This organization and the people within our cystinosis community are “family” to us and one of the greatest blessings in my life. I thank everyone for the continued love and support that has nurtured our family on our continuing journey.

During the months of April and May CRN was chosen to be the recipient of an ongoing fundraiser sponsored by Ricardo, Inc., Carl Schleuder’s employer. Donations are made for the privilege of wearing jeans on Fridays. The amount raised is then matched by Ricardo and sent to the selected charity. Thank you, to everyone at Ricardo for selecting CRN to support and raising $748. Your generosity is much appreciated.
Finance Update

The Cystinosis Research Network, Inc
Financial Review - Accrual Basis

By Jenni Sexstone

For the 3 months ended March 31, 2018

Revenues
For the three months ended March 31, 2018, total income collected of $40,000 was approximately 15% higher than the same period in 2017 from fundraising activities.

Expenses
Total operating expenses of $33,000 were less than operating expenses for the same period during 2017 of $152,000. Research grant expenditures in the first quarter of $115,000 was the primary reason for the decrease in operating expenses as compared to the same period in 2017. Excluding the grant payments made in 2017, operating expenses were 11% less, or ($3,000), than the same reporting period in 2017 due to sponsorship of a breakout session in 2017 at the Pediatric Academic Society annual conference that did not repeat in 2018. Expenses related to members of the cystinosis community participating in Rare Disease Week activities in Washington D.C. were the primary expenditures during the quarter.

CRN had net operating income of $6,000 for the three months ending March 31, 2018. Continuous fundraising activities and generous corporate support continues to provide cash resources to increase patient advocacy and family support activities in 2018 and beyond to support the cystinosis community.

Cash on hand at March 31, 2018 was $196,000. Net change in cash for the first quarter 2018 was a decrease of ($2,000). The decrease approximates the amount of net income for the first quarter less year-end 2017 accounts payable.
Update from Mexico

By Victor Gomez

Highlights from the Cystinosis Organization Mexico

• We are glad to share with all the cystinosis community that Recordati Rare Diseases is now providing Cystagon in Mexico. It took a long time and hard work to achieve this amazing goal. Now the government health services in Mexico is ready to set up a program for the care of patients with rare diseases and treat cystinosis patients in Mexico.

• In the next few months we are preparing for the first Cystinosis Nephrology Experts Training Course in Mexico City.

• Last December 2017, Victor Gomez, President of the Cystinosis Organization Mexico received the Rare Disease dedication award for caring of all cystinosis patients in Mexico, Congratulations to Victor.

Thank you!! For Fundraisers and Donations, Large and Small, CRN is Grateful

The Cystinosis Research Network wishes to thank all who have held Facebook fundraisers or other fundraising events over the past year, and for those generous folks who supported these efforts. Everything helps to support our mission of family support, education and research.

We wish to thank individuals, such as Nicole Murphy, who designated CRN as recipient of a portion of an academic scholarship she recently received. The daughter of a cousin of Marybeth Krummenacker she has always shown interest, support and compassion to the cystinosis community.

We wish to thank Betsy Dearwester whose involvement with CRN led to generous donation from The Cardinal Health Foundation.

Thank you to all those generous, caring individuals who have donated the Cystinosis Research Network in 2017 and 2018. Many are listed in our 2017 Donor Honor Roll on pages 64-70.

For any volunteer organization to survive and thrive it takes many hands, hearts and dollars. We are very grateful to you all.
Development Update

CRN Development – The Art of Giving
By José T. Morales, Vice President of Development

Seldom does a day go by that I do not receive a call from an organization requesting support in one form or another. The majority of the requests are for organizations and missions that are credible and just. The requests cover the full spectrum: American Cancer Society, Doctor’s Without Borders, Boys & Girls Club, The United Way, Local Civil Service Departments, Higher Education Institutions, etc., etc.

With our changing economic times, everyone is asking for help in some form. There are as many reasons to “give” as there are ways to give. Some people give for recognition. They want their names in programs, on park benches and in hospital lobbies. Others give expecting to be paid back or “thanked”; they give with the expectation to receive something in return. Some people want a hands-on-experience and are generous with their time. Some like to give anonymously and don’t want any recognition at all.

So why do you give? We all do in some form or another. Some of us give of our talents. Others give of themselves. We give.

I am always amazed with our community’s capacity to give. So why do we give? I am certain the reasons are as plentiful as there is stars. The most important point is, we give.

In this newsletter there are a number of giving efforts:
• Clint Moore’s 57 Mile Walk in recognition of Cystinosis Awareness Day, May 7th and other CAD fundraising events held in honor of the day
• Jack Greeley's letter campaign requesting donations to CRN in celebration of his birthday

Velyna and José Morales.
I know there are many things that bond us together. One of the strongest is our connection to this rare disorder, Cystinosis. The challenges associated with navigating one’s life with a rare disorder are significant. It demands a constant push-pull. We must rally against limitations in the medical field, and challenge scientists to be creative and relentless in their pursuit of treatments and cures. As advocates, we push lawmakers to recognize the importance of prioritizing the needs and rights of affected individuals. We pull together to create avenues for our children and adults to build community and speak their voices. Each aspect of working to prevent, combat, and treat cystinosis has one thing in common, that is raising funds. Every bit of the work we do requires funds. Supporting research to pursue a cure, building a community of experts so that when a major breakthrough in medicine is achieved, someone is there to bridge it to us. The challenges of fundraising may unfortunately always exist. Working within a niche like rare disease creates even further complexities and limitations. However, our passion for our children keeps us committed, and the strength of the community keeps us motivated to work tirelessly toward our mission to push toward finding a cure and improving the quality of life for our children.

If you are inspired to be a part of the movement for better treatments and a cure for Cystinosis, please give, no gift is too small as every contribution, you give, can be the dollar that funds the 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.
IPNA Supports Cystinosis in Africa and the Middle East

By Neveen A Soliman
Professor of Pediatrics, Cairo University
Director of the Egyptian Group for Orphan Renal Diseases (EGORD)

The first Inherited Kidney Diseases-International Pediatric Nephrology Association Teaching Course in Africa and Middle East (IKiD-IPNA 2018) was held on February 15-16, 2018 in Cairo, Egypt in collaboration with the Egyptian Group for Orphan Renal Diseases (EGORD), the African Pediatric Nephrology Association (AFPNA), African Inherited Diseases working group (AfrInKiD), and the Global Kidney Academy (GKA). Prof. Neveen A Soliman was the Course Director with an active organizing committee including Assistant Prof. Marwa Nabhan, Dr. Rasha Helmy and Ms. Islam Salama.

A dynamic and engaging 2 day course program was put together including 9 sessions two of which were dedicated to cystinosis. The main course objectives were: i) promote awareness and provide updated knowledge on inherited kidney diseases in Africa and Middle East; ii) highlight diagnostic dilemmas and therapeutic challenges in the region; and iii) foster the liaison between IPNA experts and their African and Middle Eastern counterparts to exchange clinical acumen, knowledge and expertise.

IPNA high profile experts participated in this unique regional teaching course including: Prof. Pierre Cochat, Prof. Francesco Emma, Prof. Friedhelm Hildebrandt, Prof. Elena Levchenko, Prof. Chebl Mourani, Prof. Moin Saleem, Prof. Ihab Shaheen as well as an expert ophthalmologist Prof. Omar Fakhouri in addition to 14 more expert regional speakers.

Several topics were tackled during the two cystinosis sessions: mechanisms and treatment beyond cysteine depletion, metabolic bone disease in cystinosis, growth assessment and role of growth hormone treatment, the expanding mutational spectrum of cystinosis in Egypt, management of ocular involvement, hematological manifestations and bone marrow involvement as well as peculiar cardiovascular and pulmonary phenotypes in cystinosis.

In addition to the scientific professional sessions an entertainment and art contest was
held in parallel for children. Speakers and participants joined patients and families in support and advocacy.

Huge thank to IPNA for supporting inherited kidney diseases in Africa and Middle East and to all distinguished speakers, participants, patients and families, organizers, Kasr Al ainy medical students who volunteered to arrange the children entertainment and art contest, and professional photographers Ms. Gazebya Elhamamsy and Mr. Mahmoud Abdelhay who volunteered to photograph the children entertainment event.

Finally, cystinosis is in the heart of Africa and Middle East.
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Breaking News: Recordati Acquires International Rights to Cystagon® from Mylan

Milan, 9 April 2018 - Recordati announces the completion of an agreement with Mylan for the acquisition of the rights to Cystagon® (cysteamine bitartrate), indicated for the treatment of proven nephropathic cystinosis in children and adults, for certain territories, including Europe. The product was previously commercialized by Orphan Europe (a Recordati group company) under license from Mylan.

"We are very satisfied with the agreement reached with Mylan for the acquisition of the rights to Cystagon®", declared Andrea Recordati, CEO and Vice Chairman. "Our rare disease company Orphan Europe has, over the years, successfully made this product available to patients suffering from nephropathic cystinosis in Europe and in many other parts of the world. The definitive acquisition of the rights enables us to continue offering this life-saving treatment to patients".

Recordati, established in 1926, is an international pharmaceutical group, listed on the Italian Stock Exchange (Reuters RECI.MI, Bloomberg REC IM, ISIN IT 0003828271), with a total staff of more than 4,100, dedicated to the research, development, manufacturing and marketing of pharmaceuticals. Headquartered in Milan, Italy, Recordati has operations in the main European countries, in Russia, other Central and Eastern European countries, Turkey, North Africa, the United States of America, Canada, Mexico and in some South American countries. An efficient field force of medical representatives promotes a wide range of innovative pharmaceuticals, both proprietary and under license, in a number of therapeutic areas including a specialized business dedicated to treatments for rare diseases. Recordati is a partner of choice for new product licenses for its territories. Recordati is committed to the research and development of new specialties with a focus on treatments for rare diseases. Consolidated revenue for 2017 was € 1,288.1 million, operating income was € 406.5 million and net income was € 288.8 million.

For further information:
Recordati website: www.recordati.com

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

Visit https://www.cystinosis.org/family-support/in-your-area

Send your contact info to the e-mail listed to learn who is in your area. It’s that easy!
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 website www.cystinosis.org, the popular online Cystinosis Facebook Support Groups, 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
Join the Cystinosis Research Network Today!

<table>
<thead>
<tr>
<th>Category</th>
<th>Fee</th>
</tr>
</thead>
<tbody>
<tr>
<td>Immediate Family</td>
<td>$20.00</td>
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<tr>
<td>Extended Family/Friend</td>
<td>$25.00</td>
</tr>
<tr>
<td>Professional</td>
<td>$35.00</td>
</tr>
</tbody>
</table>

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

<table>
<thead>
<tr>
<th>Agency Name</th>
<th>The Cystinosis Research Network, Inc.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Non-Profit Tax ID #</td>
<td>04-3323789</td>
</tr>
<tr>
<td>Address</td>
<td>302 Whytegate Ct., Lake Forest, IL 60045</td>
</tr>
<tr>
<td>Telephone</td>
<td>1-866-276-3669 (toll free), 1-847-735-0471</td>
</tr>
<tr>
<td>Fax</td>
<td>847-235-2773</td>
</tr>
<tr>
<td>E-mail Address</td>
<td><a href="mailto:CRN@cystinosis.org">CRN@cystinosis.org</a></td>
</tr>
<tr>
<td>Web Page</td>
<td><a href="http://www.cystinosis.org">www.cystinosis.org</a></td>
</tr>
</tbody>
</table>

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!

[MissionFish](https://org.amazon.com/npo/portal/marketing-tools/ref=org_prt_gwh_mt)

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

**CRN Vision and Mission**

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

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