Cystinosis Research Network Meet-ups

This year the Cystinosis Research Network (CRN) hosted three cystinosis meet-ups in Houston, Indianapolis, and Charleston. Houston’s guest speaker was Dr. Ellenberg, Indianapolis’ guest speaker was Dr. Gipson, and Charleston’s guest speaker was Dr. Nesterova. All three doctors gave an excellent explanation of cystinosis along with their expert advice and opinions on various topics. They answered many questions and listened to many concerns from attendees. An absolute outstanding job was done by all three of these doctors.

Carrie Ostrea also presented at the first two and gave a talk about advocating for yourself as well as how to get involved. Carrie is a wonderful asset with her experience and knowledge as she also runs her own nonprofit in memory of her daughter.

Maya Doyle presented at the final meeting with her talk about coping

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The President’s Letter

As the president of the Cystinosis Research Network, I am the one who often times gets the credit for all that is accomplished within the network. I get calls, texts, and messages from community members all the time thanking me for all that the CRN does for this community, and many times their thanks is pointed toward me. As much as I do appreciate this outreach and words of encouragement, I am not always the proper one to thank. I recently sat in a room with two other people to plan and finalize the details of our new upcoming website. I sat there all day and tried to give my opinion many times, but to be honest, the technical stuff is a bit above my intelligence level. So most of the day I listened, learned, and was impressed at the amount of detail and work that goes into something like that. My point is that I am only one person in a network of many. Nothing would be possible without the many volunteers that give their time to help make this organization an extreme success. Everyone on the executive committee and on the board has his or her own strengths and weaknesses. I am no exception. I wish I had a list of how many hours and sacrifices this board makes on a weekly basis. It certainly does not go unnoticed or unappreciated. I cannot thank this team of people enough for all that they do.

One thing that this wonderful team of people has in common is our focus. That focus is this community. Whether newly diagnosed, adult veteran, parent, sibling, caretaker, or medical professional, this network supports you and has your best interest in mind. This focus never changes. Why should it?

Regardless of what you’re up against, the CRN is here to help. Reach out to any of our board members, and we will get you the help you need or at least point you in the right direction. We have many resources available and many people with all different types of backgrounds willing to help. We are just a phone call, text, or email away.

Be well, stay strong, and Happy Holidays!

Your President,
Clinton Moore
Cystinosis Research Network Meet-ups
Continued from Page 1

mechanisms and how to recognize and handle stress or depression that often comes along with rare diseases. She offered many available resources for our community.

I also presented at all three meeting with a talk describing CRN and our current projects. I also touched on involvement and advocating for yourself.

Each cystinosis meetup was a huge success with great feedback from every attendee. Families talked and shared stories with each other both before and after the events. Watching the families interact has always been one of my favorite parts of any type of cystinosis gathering. I believe we often learn more from each other than we do from any textbook.

Thank you to all who were involved in making these meetings happen and a very special thank you to all those who attended. I look forward to hosting more of these events soon.

The Cystinosis Research Network is happy to congratulate the 2018 recipients of the Sierra Woodward Sibling and Individual with Cystinosis Scholarship Awards. There were many applications this year, making it difficult to choose our winners. We want to thank all those who applied. To those who were not selected this academic year, we encourage you to reapply next year.

Congratulations to the recipients of the Individual with Cystinosis awards, Kathleen Roberts and Jack Kitchens!

Kathleen Roberts plans to attend the University of British Columbia at Okanagan’s in nursing program. Her dream and goal in life is to build an excellent standard of supportive health care for all.

Jack Kitchens plans to study biology at the University of Georgia. Jack’s time in Cozumel scuba diving and experiencing the underwater sea life has led him to further his education in the area of marine biology.

The recipients of the Sierra Woodward Scholarship for siblings of individuals with Cystinosis are Rhyana Christine Juutilainen and Emily Ann Bogan.

Rhyana Christine Juutilainen will attend the University of Nebraska at Omaha, where she will study Criminology. She wants to learn more about the minds of individuals who commit criminal acts.

Emily Ann Bogan plans to attend Central Community College in North Carolina and major in Criminal Justice Technology. After receiving her Associate Degree she wants to pursue a Bachelor’s Degree. Upon completion, her goal is to work with individuals struggling with addiction and/or youth who are at risk.

Thank you again to all who applied and remember you can always apply again. Look for the scholarship application announcement in our spring newsletter.
CRN Attends European Cystinosis Conference in Berlin, Germany

By Christy Greeley, VP of Research and Executive Director

Christy Greeley, Executive Director/VP, Research and Jen Wyman, VP, Family Support, were pleased to attend the 2018 Cystinosis Network Europe conference on behalf of CRN and the US cystinosis community. The meeting was held in Berlin, Germany July 13-15. The program included a meeting of the E-Rare Consortium where novel therapies for cystinosis were presented. Included were projects on The zebrafish model of cystinosis, new candidate molecules for cystinosis treatment, cystine levels and metabolomics analysis in cystinotic cells, effects of genistein on cystinotic mice, kidney regeneration in cystinosis, priming nephron progenitor cells isolated from fetal urine, bone defects in cystinotic mice, and novel biomarkers of cystinosis.

International speakers were on hand to present information to the families and professionals in attendance on the basics of cystinosis, system specific information, a vision for the future of cystinosis treatment, plus many small group discussions on a variety of topics. In addition, a Poster Session took place which included plenary presentations by each author.

Jen and Christy were also pleased to connect with cystinosis advocacy leaders from around the world to share progress and best practices, as well as attend a presentation by Eurodis, the European rare disease blanket organization, on the institution of Community Advisory Boards in which groups of patients offer their expertise to public or private sponsors of clinical research, so important in particular in the area of rare disease.

Thanks to the organizing committee for including the Cystinosis Research Network and congratulations on a most informative meeting. We look forward to our international colleagues joining us next July for the 2019 CRN Family Conference.
THANK YOU!
This meeting was held in Presidente Intercontinental Hotel Mexico City on **June 12 & 13, 2018**. There are around 18 cystinosis patients in Mexico city, as often happening in other countries, cystinosis as a rare disease makes it difficult to have physicians knowledgeable about the diagnosis and treatment.

**The Cystinosis Organization Mexico, Craig Langman MD** and **Elena Levchenko MD** with the **sponsorship of Horizon Pharma Pharmaceuticals, Inc** decided to organize this great meeting, having in mind the idea of getting together nephrologists who actually are taking care of patients with cystinosis today.

Elena and Craig created a great agenda which focused on the following main topics:

- **Nephropathic Cystinosis Types and natural history of untreated patients with cystinosis.**
- **Cystine reduction therapy.**
- **Dialysis and transplantation.**

All this including a web-based Case-Based Pre-Test and a conclusion post-test which was handled by Benjamin Joslin, Clinical Research Project Manager for Dr Langman.

With all this together those nephrologist had a busy learning day on cystinosis, we appreciate their attendance. Cystinosis Organization Mexico Will still be organizing such this meetings to raise awareness on cystinosis as part of their principal objetives.
Ireland Updates

Cystinosis Ireland has been working hard on both our advocacy and research programmes this summer – we were waiting for the often-cited quiet time, but it never came!

Our advocacy work focused on an educational briefing for politicians in our parliament in late September. Organised by Cystinosis Ireland, people living with rare conditions and their advocates shared their first-hand experience of living with their condition with our TDs (elected representatives) and Senators. We also used sessions to raise issues which need urgent political support. We had a strong response from the politicians who attended, and we look forward to their support for our community’s needs.

Our research programme is continuing steadily, and we are keeping abreast of new developments in the research arena as they are occurring. We were delighted to see research we are part-funding in New Zealand getting an excellent write up, highlighting the revolutionary way human kidney tissue is grown from stem cells. The full article can be found here: bit.ly/2z6pF2q.

Also on our research agenda is the Fifth Dublin Cystinosis Workshop which will take place on 26 and 27 April 2019, and the Cystinosis Network Europe conference which it has been confirmed will be hosted in Dublin in the summer of 2020. We look forward to welcoming friends and colleagues to share their knowledge and expertise with others in the research community and with the families who attend these conferences. The 2020 meeting will build on the successes of the Berlin meeting earlier this year, and the work we expect to emerge from the Dublin Cystinosis Workshop in 2019.

Everyone with an interest in research should keep in touch with research@cystinosis.ie and our website for details of the Seedcorn (in collaboration with Cystinosis Research Network and Cystinosis Foundation UK) and other funding opportunities.

Finally, our advocacy and research priorities are coinciding in the plan to develop a Community Advisory Board (CAB) for Cystinosis, under the auspices of the Cystinosis Network Europe. The CAB process has been formalised by Eurordis (the European Rare Disorders Organisation) in recent years following the significant successes HIV patients and their advocates achieved through these processes in the 1980s. The CAB creates a formal structure for patient organisations to interact with pharmaceutical and health technology companies to help guide and support the companies’ research agendas to meet the needs of patients. The processes are thoroughly supported with training for the patient advocates and strict rules to guide the interactions. A hugely important part of the CAB is that each meeting is patient led with the agenda being set by them. We envisage a lot of work in the coming months to develop our CAB process but look forward to useful and productive engagement as a result of it.
Around Christmas 2017, we felt in our hearts there was something wrong with our son when he began to not act like his usual self. We tried searching of the cause behind these changes but kept hitting dead ends. It wasn’t until April 25th, 2018 when our lives changed forever.

Our son, Benson, was born March 16th, 2017. He was an all-around healthy baby. We couldn’t have asked for anything better. Or so we thought. When his 1st birthday approached is the time everything started going downhill. He wouldn’t eat anything by mouth other than breastmilk without vomiting. We tried time and time again to get him to eat solid food thinking it was something as simple as a texture issue, but we never succeeded. Along with his lack of eating he showed signs of excessive thirst and excessive urination and began losing weight. The weight loss became a concern when he lost four pounds in one month classifying him as failure to thrive.

On April 18th, 2018 Benson came down with the stomach bug and he struggled to come back from it. He continued to act lethargic, his eyes were sunken in and we became really concerned. We took him to the doctors and they sent us to get blood work done to check his levels. That following morning, April 24th, we received a phone calling telling us that Benson’s potassium levels were dangerously low and that we needed to head to the emergency room. Our local hospital was unable to give him the proper level of care, so we were transferred to a children’s hospital an hour away and that is where we spent the next 28 days.

Benson’s hospital stay still haunts me to this day. Seeing him in pain and terrified of the doctors and nurses and not being able to do anything to help him was the hardest part. The first evening at the children’s hospital consisted of potassium supplements intravenously to try and get his levels up to where they needed to be. The second day we saw a handful of specialists to try and rule out or determine possible diagnoses. By the end of the second day, April 25th, myself and Benson’s dad were sat down by one of the many doctors and were told that Benson has Fanconi Syndrome and a possible diagnosis of cystinosis. Having someone tell us that our sweet baby has a rare incurable genetic disease was heartbreaking. We felt like our world was falling apart. The doctors were unable to give a definitive diagnosis because his blood work had to be sent to a lab in California but they were certain that was what he had. The weeks following were some of the hardest on Benson and myself. He had his blood drawn every four hours to monitor his levels even throughout the night. He was exhausted and scared. As the blood draws continued he became harder and harder to stick so the nurses had to move to his hands, feet, and head. He was covered in bruises from head to toe and was so sore. After about four days of the constant blood draws, Benson was finally able to get a PICC line to make checking levels a lot easier. The
next step was to have a g-tube put in to help him gain some much-needed weight as well as administer meds. 24 hours after the g-tube was put in we were able to start feeds to get a higher number of calories into him aside from my breastmilk. During the hospital stay my three-year-old daughter Lillian struggled emotionally. She had never been away from me more than one day until Benson was admitted. She had a hard time understanding why she couldn’t stay or why we couldn’t go with her. Seeing the hurt in her eyes every time she would leave the hospital broke my heart.

Everything remained the same over the next few weeks focusing on keeping Benson stable. On May 13, 2018 (Mother’s Day) we were told that Benson in fact has cystinosis. Now with a definitive diagnosis, the doctors were able to start the meds needed to slow down this disease and the damage done to the kidneys. It was nine days after the diagnosis that we were discharged from the children’s hospital and were finally able to return home.

When we returned home, we all had an adjustment period. We had to shift our lives around to accommodate for his medication and feed schedules. We had to find a new “normal.” One of the biggest challenges about returning home was Lillian had a hard time understanding why she again was receiving less attention compared to Benson. We did the best we could to make her not feel less than and in time things did get better. Now she is excited to help with giving Benson his medication or will grab the vomit bag when he is about to get sick from one of his feeds.

We are six months into this journey and Benson shows small improves every day. He currently receives 10 meds every six hours along with feeds for a total of 18 hours a day. He has gained 11 pounds and recently began walking which are both big steps in the right direction. We do struggle with his levels because they fluctuate quite a bit, but meds are constantly adjusted to try and manage that. He now receives early intervention (speech, OT, and PT) to help him catch up to where he needs to be. Benson is back to his happy self and LOVES to play with his sister. It will continue to be a challenge every day and I’m sure we will hit bumps in the road, but all we can do is keep fighting and praying for a cure. Until that day comes, this life is our new “normal.” Benson is a warrior.
Cystinosis Only Makes Me Stronger

By Megan Morrill

Growing up my parents always told me I was an extraordinary person living an ordinary life. As a young child I never quite understood what this statement meant or how it related to me, but as I’ve gotten older I have a whole new understanding of what this means. Going into my senior year at Albion College I’ve been reminiscing about the past years and the experiences I’ve had. I’ve had the opportunity to do so many phenomenal things in my college years including preparing for a career in Occupational Therapy, studying abroad, and having the privilege of training my horse Amigo from start to finish. Before I explain all these great opportunities I’ve had, I want to tell you a little bit about how I got to where I am today.

When I was a child I never considered myself different from everyone else. I went to school like every child my age, had sleepovers like every young girl, and did as many sports as I possibly could. What I didn’t know at the time was not everyone’s mom came to school at lunch in order to give their child medication. I didn’t realize it was not normal for parents to also spend the night at their child’s sleepover in order to set up and monitor the child’s feeding pump all night. Or that most parents did not constantly carry around multiple bottles of water and snacks at sporting events just in case their child got fatigued or dehydrated. To me all these things were just normal parts of my life and I had no clue other children’s parents didn’t do these things.

Looking back at my childhood I see this as a big factor to where I am today. My parents made it their priority to let me do anything I wanted and not let cystinosis hold me back. Yes, there had to be some modifications to activities when I did them, but my parents always willingly gave up their time to make me feel like every other kid instead of taking the easy way out and saying no. I believe this is a big contributor to my independence and ability to conquer even the biggest task. So thank you Mom and Dad for allowing me to live the most ordinary yet extraordinary childhood possible.

When I started college I had no idea what I wanted to go into. Originally I thought dentistry would be a good career path for me, but I ended up deciding that was not the right fit. Finally, I settled on Occupational Therapy and knew it was the perfect career path for me. During the summer of my freshman year I shadowed an occupational therapist, Courtney Sumpter, at Northern Michigan Equine Therapy and absolutely fell in love with the work. Courtney worked with a very diverse group of children and adults with Cerebral Palsy, Pitt Hopkins, Autism, brain injury’s, etc. The more I observed and worked with the clients I found myself forming a strong connection with these individuals. The struggles I went through, and still go through, with cystinosis were very similar to the struggles these
individuals go through on a daily basis. After this experience I knew I wanted to make an impact and help individuals live their best lives possible. Hopefully in a couple years I will be a certified Occupational Therapist and be able to impact individuals lives in a positive way.

Reminiscing on my college career the of the most memorable experience was studying abroad in Dublin, Ireland for a semester. During these four months I took classes while also having the opportunity to intern at an outpatient clinic for patients with traumatic brain injuries. Getting to immerse myself in another culture and experience the healthcare system of another country was truly an irreplaceable experience. I met such amazing people that I am still friends with today and learned a tremendous amount about myself. And of course on my time off the other students and I traveled all around Ireland and to other countries including Norway and England. These experiences were truly irreplaceable and I’m so thankful for this opportunity.

Another memorable experience during my college career is training my horse Amigo. I started riding horses at age six and once I took my first riding lesson there was no turning back. I was hooked from day one. There was something special about having the ability to tell a thousand-pound animal what to do and have them actually listen. The summer of 2017 Amigo came into my life as an untrained 14-year-old mare. The minute I started working with her I absolutely fell in love. Over this past year and a half there have been many ups and downs (including hitting the hard arena footing a few times) but it has been the most rewarding experience in my lifetime. The countless hours at the barn and hundreds of hours spent in the saddle are all worth it when you finally reach the goal you’ve been striving to achieve for months.

All these experiences in my life have shaped me into the person I am today. I now understand what my parents meant when they told me I am an extraordinary person living an ordinary life. Cystinosis gives me many struggles most people do not have to live with, but I do not let these struggles stop me from doing the things I love. Like we say in the equestrian world “when you fall off your horse, you have to get right back in the saddle.” I do not let the struggles of cystinosis define me, but see it as something that can only build me up and make me a stronger person.
A Financial Safety Net for Cystinosis Patients

For many cystinosis patients, an unexpected diagnosis is often followed by the realization that your healthcare coverage and savings are not enough for the out-of-pocket costs that are ahead. That scenario was no different for Jodi and her husband Alan, when they found out their son Malachi was diagnosed. Thankfully, the national non-profit charitable organization Good Days was here to help.

At just nine months old, Malachi was having symptoms that required he be checked into a hospital for 10 days. There, Jodi learned of his diagnosis and was given information about Good Days.

“It was a relief to have a diagnosis,” says Jodi, but treatment and next steps would be a lot to process. “I now thank the Lord that Good Days was there because I really don’t know how I would have done this without them.”

Malachi is doing much better today and is now a shy, artsy five-year old who loves to draw, do puzzles and listen to music. Good Days helps with the cost of his treatment and travel expenses.

Good Days supports many cystinosis patients across the country by assisting with their co-pay costs, allowing families to receive treatment without destroying their finances. In some cases, Good Days also provides additional financial assistance for health insurance premiums and treatment-related travel expenses. Since 2003, Good Days has provided more than 800,000 grants and helped more than 500,000 people with access to healthcare resources.

To find out if you qualify for financial assistance call (877) 968-7233 or visit mygooddays.org.

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.com.
Symbol of Eternal Support: Father Gets Tattoos of Son’s Scars

Living with cystinosis has earned Chandler Moore a few battle wounds. After mulling over a way to symbolize the journey he’s sharing with his son, Clinton decided to permanently tattoo his son’s scars on his own midsection.

Q: Can you explain what caused the scars we see in this picture?
A: The upper scar is from the placement of a g-tube. This was needed to give extra nutrition and administer meds. The lower scar is from a more major surgery to create something called a mitrofanoff. Simply put, it’s a channel to insert a catheter from his belly button to his bladder to help with drainage.

Q: Are these your only tattoos?
A: Yes. I’ve never had an interest in tattoos... and still don’t. But after years of wishing I could take this disease for Chandler knowing I couldn’t I realized this is a way to share it. If he has to be “imperfect” so can I. Every scar that this disease gives to him my tattoo artist will give to me. I think this has impacted me personally more than it did him. Now I feel like I’m carrying it [cystinosis] with him.

Q: What did you son, Chandler, think of this idea?
A: Chandler thought it was a great idea and was quite happy to see me permanently tattoo them [scars] on me. He was even asked to attend on tattoo day so the artist could use him as a model.

Q: What do you hope to accomplish as a result of your new ink?
A: My hope was to show my son that he is not in this alone. I’ve got him. I always will. There is nothing I won’t do for him and if I could take this disease for him I would do it in a second. I would take it for every patient if I could. Also, to show him there is nothing wrong with being a little “imperfect”. Most imperfections are absolutely perfect.
You are invited to the City of Brotherly (and Cysta-ly) Love!

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, the only waterfront hotel at the doorstep of the city’s center. 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, 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 Kitchen. Walk to Philadelphia attractions including the Liberty Bell, Independence Hall, Museum of the American Revolution and Blue Cross Riverrink Summerfest.

For more information on the hotel please visit their website at bit.ly/2QtIRh4.

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.

Don’t miss out on one of the largest gatherings of individuals and families with cystinosis in the WORLD. Visit our website in 2019 for online registration at cystinosis.org.
Family Support Update
By Jen Wyman, Vice President of Family Support

Each time we print a newsletter (precisely twice a year), I am reminded of how such a small community of patients has such an incredible wealth of support from family and friends, both in the way of monetary donations and emotional support. I have served, in some capacity, on the board since Kacy’s diagnosis 12 years ago. My husband, Tim, stumbled across the CRN website looking for information about cystinosis while Kacy was still in the hospital. By the time she was released two weeks later, we had been in contact with the hard working volunteers who make CRN work -- the same people who are parents of cystinosis patients or patients themselves, the same people who we call extended family and friends today, the same people who understand completely what you have gone through in your journey. These people needed to be our people, so we got involved. Within the first year, we held our first of a dozen or so fundraisers (raising over $60,000), attended our first conference, and I took a position on the board.

I have served as VP of Development and currently serve as VP of Family Support. I have seen first-hand how generous people can be. You open your heart and share the journey, and people open their wallets and they give over and over and over again. I see it repeatedly in our community.

Serving on the Development side of CRN gave me the understanding of how important fundraising is. The money we raise funds the research we need to make the lives of our loved ones better. It’s important. It’s necessary. It keeps our organization alive. But it’s only part of our purpose. It’s been good for me to serve in different capacities. I have been able to see how committees serve different purposes while attending to the same goal. Supporting our families -- all of them with their variety of differing needs and issues that arise with a rare disease is what CRN is all about. Our stories are the same, but different. We are in a collective fight to keep our heads up and our hearts full, our kids healthy and our families happy. Those who are on a good stretch lift those that are facing obstacles. We use social media to make our world smaller and make our families feel heard. We connect people to each other within our own country and beyond. I have been honored and humbled to serve in this position, to be “here” for you and to “hear” you.

“One of the most vital ways we sustain ourselves is by building communities of resistance, places where we know we are not alone.” – Bell Hooks

Thank you to our TEAM KACY supporters. Many of you who have been with us since the beginning of our cystinosis fight. Our last letter campaign raised over $12,000. Your ongoing contributions and your continued support of our family keeps our hope alive. Much Love!!
Meschke Family Fundraiser

By Cheryl Meschke

On September 30th, 2018 the Meschke Family along with Shady’s Tap Room in Brooklyn Michigan hosted “A Night for Jax”. The event was such an amazing success, outdoing any expectations we had! With the amazing support from the community, our friends and family the event raised $10,221 all for the Cystinosis Research Network! It was the perfect day with 50/50 raffles, a huge silent auction, great food, drinks, and live music! We were surrounded by love and couldn’t be more thankful for the immense support we have had, not just at this event but for our whole family since our son’s diagnosis. As we continue our personal fight it’s a blessing knowing we have such a support behind us. We are truly thankful.

Arnold Foundation Supports CRN Mission

By Jeff Larimore

Earlier this year, the Cystinosis Research Network was elated to receive a contribution of $15,000 from the Norman J. and Gerry Sue Arnold Foundation of Columbia, SC. This generous donation was made in the honor of Sarah Larimore, who had just endured her first kidney transplant shortly before the receipt of the award. Sarah commented that “it was really exciting for CRN to receive this wonderful gift so that we can keep moving forward in finding a cure for cystinosis”.

Over the past three decades, the Arnold Foundation’s philanthropy efforts have concentrated on improving overall public health and wellness. Past charitable donations include an endowment to support the teaching and research efforts in Public Health at the University of South Carolina. It has also supported over 100 doctoral students as Norman J. Arnold Doctoral Fellows in Public Health.

More recently, the Arnold Foundation broadened their commitment to public health with the creation of the Gerry Sue and Norman J. Arnold Institute on Aging. The mission of this program is to promote healthy aging by addressing issues in early childhood, nutrition and food safety along with heart and mental health for an aging population.

Jay Swearingen, a trustee with the Arnold Foundation, added, “The Foundation is happy to support the Cystinosis Research Network and its mission. We are interested in improving the quality of life for cystinosis patients and working for a long-term solution for all who suffer with this rare disease.”
#GivingTuesday: Facebook and PayPal Pledge $7 million to U.S. NonProfits

Facebook has created a simplified approach to online fundraising that can reach “friends” in a matter of moments. Their tools automatically send deadline reminders and keep followers updated on how they are tracking against their goals. This year, Facebook took the generosity associated with #GivingTuesday one step further by teaming up with PayPal. The duo committed to match $7 million in Facebook donations made to U.S. nonprofits on November 27th, 2018. The match started at 8:00 a.m. Eastern and the company formally announced the match limit was reached before 9:05 a.m. Eastern.

YOUR IMPACT
Since 2015, the CRN has been a registered partner of #GivingTuesday. Through this partnership, we’ve been able to raise thousands of dollars that will help researchers improve treatments and continue forward progress towards finding a cure.

A big THANK YOU to those who submitted a #MyGivingStory which gives CRN a chance to win $10,000 by sharing what/who inspires you to give. We are also grateful for your support of CRN’s #GivingTuesday campaign through shares on social media and direct donations. We are humbled by the individuals that choose to honor our organization with personal fundraisers during #GivingTuesday and throughout the year.

OTHER WAYS TO GET INVOLVED
If fundraising isn’t for you, there are other ways to help the community. Volunteering your time, lending a skill and becoming a voice for cystinosis are among a few options. Learn more at cystinosis.org/how-to-help/get-involved or email info@cystinosis.org.

WHAT IS #GIVINGTUESDAY?
#GivingTuesday is a global day of giving fueled by social media and collaboration. It is celebrated on the Tuesday after U.S. Thanksgiving and kicks off the end-of-year charitable giving.

Next year’s event will be held on December 3rd.
The Cystinosis Research Network, Inc. Financial Review

By Jenni Sexstone, Treasurer

For the 9 months ended September 30, 2018:

Revenues
For the nine months ended September 30, 2018, total income collected of $388,000 was approximately 22% higher than the same period in 2017 from fundraising activities.

Expenses
Total operating expenses of $257,000 were less than operating expenses for the same period during 2017 of $564,000. The primary reason for the decrease in expense is prior year expense for the 2017 CRN Family Conference. Research grant expenditures for the period were $123,000 compared to $215,000 in the same period for 2017. Excluding the grant payments and family conference expenses, operating expenses were $131,000 compared to $97,000 in the prior year due to increased advocacy at Rare Disease Week, attendance at the World Conference Patient Adherence & Engagement Summit, the recent Cystinosis Patient Meetups as well as website and branding updates.

CRN had net operating income of $132,000 for the nine months ending September 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 September 30, 2018 was $300,000. Net change in cash for the first nine months of 2018 was an increase of $103,000.

Designate Cystinosis Research Network on Amazon Smile

Support our organization while you do your holiday shopping on Amazon! Designate CRN as your favorite charity on Amazon and 0.5% will come back to the cystinosis community.

How do you do it?
1. Visit smile.amazon.com
2. Select your account
3. Under “Settings” select “Change Your Charity”
4. Type and select “Cystinosis Research Network”
Hello Cystinosis Community! I am Herberth Sigler, and this is my first time writing in the newsletter. First, I would like to say I am honored to be part of the CRN family and being the VP of Development for this term. Three months ago when I accepted the invitation from the executive board, but the desire to serve and help to achieve CRN goals has long been a daily activity for me. Families, friends, and doctors who guide us to keep our loved ones well-controlled despite all the challenges will continue tirelessly to find better treatments and ultimately a cure for cystinosis.

Today, I would like to say thanks to those members within as well as outside of the cystinosis community who have the willingness to donate or fundraise large or small contributions -- both of which make a difference by bringing awareness and advocacy, and inspiring people to pursue our common goals. When doing fundraisers, we help to achieve objectives, finance new research and writing scientific publications, awaken interest and educate medical professionals to understand the necessities of our loved ones in treating and caring for cystinosis patients. We move forward and with these actions, we are cementing all gained experiences from patients and doctors to bring better treatments and continue ahead to our ultimate goal – a cure.

A couple of years ago, when my family was in the early stage of being diagnosed, the whole world ended for us. Not fully comprehending what happened, the cystinosis community -- people like you that are reading this newsletter -- gave us hope. We were encouraged by parents that went through what we are going through, but ten, fifteen or 20+ years ago without drugs like cysteamine. Today, the availability to have cysteamine in a pharmacy is evidence of the importance of a community that is gathering, communicating, empowering in sharing experiences and being together in pursuing our goal.

End-of-the-year holidays are right here. Please remember us on this Giving Tuesday or throughout the holidays. You can help us anytime by donating at our website, cystinosis.org. Also, Cystinosis Research Network (CRN) can be selected as your preferred charity on AmazonSmile, just remember to log-in on AmazonSmile and a portion of qualifying purchases will be donated to the CRN.

If you have ideas or just would like to be involved in helping/volunteering in fundraiser activities, please feel free to send me an email to hsigler@cystinosis.org.

Finally, here is a picture of my family and the little one having a good time on a summer day. Every time we see the picture of our daughter laughing, we give thanks to doctors and those families that went through initiating clinical trials more 20 years ago to make it possible. Because of them, we can go to the pharmacy and get the medicine as part of our routine and then go on living our lives.

Happy Holidays!

Herberth Sigler (left) with wife Jessica and daughter Martina.
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 $200,000 and has funded $4.5 million total in research grants and fellowships. CRN funded a Cystinosis fellowship at the National Institutes of Health, 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 have focused on 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 by the end of 2018 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
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 Total Grant: $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 Levchenko (Principal Investigator)

Katholieke Universiteit Leuven, Belgium

Year Two: $88,493 Total Grant: $165,494

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 three: $100,980 Total Grant: $467,950
Travel Addendum: $38,352

The goal of this proposal continues to focus on brain-related changes in the lysosomal disease, cystinosis, through the use of complementary state-of-the-art neurocognitive studies (in cystinosis patients, Aim 1), cell biological analyses and possible treatment strategies (in the mouse model, Aims 2-3). Progress through year 02 of the proposal are outlined below.

AIM 1: To explore sensory processing and multisensory integration as potential biomarkers using high-density electrophysiological mapping techniques in individuals with cystinosis.

This section of the report details progress in the human arm of the CRN project entitled - “Mechanisms Underlying Neurocognitive Changes in Cystinosis”. Under this arm of the project, our main aim was to “explore sensory processing and multisensory integration as potential biomarkers using high-density electrophysiological mapping techniques in individuals with Cystinosis”. Therefore, initially, we recorded high-quality data from 7 patients with Cystinosis while they responded to a sensory processing task. We performed preliminary analyses of those data, compared the outcomes to similar recordings in another lysosomal disorder (Niemann-Pick-C (NPC); N=17) and to an already collected extensive normative dataset recorded from a cohort of matched neurotypical control participants (N=84). The most surprising aspect of those results was the strikingly “normal” patterns of multisensory behavior and neurophysiological responses that we obtained in Cystinosis, in stark contrast to those obtained in the NPC population. As a consequence of this finding, we decided to apply additional paradigms that tap into sensory processing and executive functions, which, based on the clinical phenotype of individuals with Cystinosis, are likely to provide sensitive brain measures of neural function/dysfunction in the Cystinosis population. In this second phase, we have collected data from 32 patients with Cystinosis and from 37 neurotypical control participants and performed analyses of part of the behavioral and the electrophysiological data. In this cumulative report, we indicate major progress over the past six months, since our last report, with blue font.

Scientific communications: We presented preliminary analyses of the data at the Cystinosis Research Network Family Conference in Utah. We also presented these data at the Pediatrics Research Day and at the Lysosomal Rounds at the Albert Einstein College of Medicine and to our Lysosomal Storage Disorders Grand Rounds at Einstein/Montefiore group (April 2018), and will present them at the International Meeting of the Psychonomic Society taking place in the Netherlands (May 2018).
have started preparing the data from two of the datasets for publication, and are currently working on the first manuscript.

Recruitment Efforts: We engaged in extensive recruitment efforts through social media and during the Cystinosis Research Network Family Conference. Furthermore, enrollment capacity was greatly increased through the addition of funds from the CRN to fly families in for two days of data collection. We have met our new recruitment targets and have completed data collection. In the past year and a half, we collected data from 32 individuals diagnosed with cystinosis (19 children, 6 adolescents, and 5 adults) and from 37 neurotypical controls (16 children, 12 adolescents, and 9 adults). We propose to collect data from 9 more adults and 8 more adolescents with Cystinosis (plus healthy age matched controls) so that we may assess how perceptual and cognitive functions change over development in Cystinosis.

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 cystinosis.org/research/article-library/cystinosis-overview.

Dr. J.J. Zaritsky joins the CRN Medical Advisory Committee

Meet Dr. J.J. Zaritsky, the newest member of the CRN Medical Advisory Committee.

From the signature bow tie to his easy-going personality, Joshua J. Zaritsky, MD, PhD has become a beloved and passionate advocate for the cystinosis community.

Serving as Pediatric Nephrology Division Chief at Nemours/A.I. duPont Hospital in Wilmington, Delaware, “Dr. J.J.” has received accolades from the National Kidney Foundation and Philadelphia Magazine’s Top Doctors list. It is a privilege to welcome him to the CRN Medical Advisory Committee!

For more information on the CRN Scientific Advisory Board and Medical Advisory Committee members, please visit the CRN website at cystinosis.org/about/leadership.
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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
National Academy of Medicine Elects 85 New Members

The National Academy of Medicine (NAM) today announced the election of 75 regular members and 10 international members during its annual meeting. Election to the Academy is considered one of the highest honors in the fields of health and medicine and recognizes individuals who have demonstrated outstanding professional achievement and commitment to service.

“This distinguished and diverse class of new members is a truly remarkable set of scholars and leaders whose impressive work has advanced science, improved health, and made the world a better place for everyone,” said National Academy of Medicine President Victor J. Dzau. “Their expertise in science, medicine, health, and policy in the U.S. and around the globe will help our organization address today’s most pressing health challenges and inform the future of health and health care. It is my privilege to welcome these esteemed individuals to the National Academy of Medicine.”

New members are elected by current members through a process that recognizes individuals who have made major contributions to the advancement of the medical sciences, health care, and public health. A diversity of talent among NAM’s membership is assured by its Articles of Organization, which stipulate that at least one-quarter of the membership is selected from fields outside the health professions — for example, from such fields as law, engineering, social sciences, and the humanities. The newly elected members bring NAM’s total membership to 2,178 and the number of international members to 159.

Established originally as the Institute of Medicine in 1970 by the National Academy of Sciences, the National Academy of Medicine addresses critical issues in health, science, medicine, and related policy and inspires positive actions across sectors. NAM works alongside the National Academy of Sciences and National Academy of Engineering to provide independent, objective analysis and advice to the nation and conduct other activities to solve complex problems and inform public policy decisions. The National Academies of Sciences, Engineering, and Medicine also encourage education and research, recognize outstanding contributions to knowledge, and increase public understanding. With their election, NAM members make a commitment to volunteer their service in National Academies activities.

This year’s newly elected members included one of our very own Medical Advisory members, Dr. Gahl.

William A. Gahl, M.D., Ph.D., senior investigator, Medical Genetics Branch, and clinical director, National Human Genome Research Institute, National Institutes of Health, Bethesda, Md.

For contributions that include creating the Undiagnosed Diseases Program within intramural NIH to meld individualized patient care with next-generation sequencing and to provide insights into new mechanisms of disease; spearheading expansion to the national Undiagnosed Diseases Network and the Undiagnosed Disease Network International; and championing the sharing of genetic databases and best practices.
Montreal – The Research Institute of the McGill University Health Centre’s (RI-MUHC) push to treat a rare genetic disease in children – with a higher percentage found in French Canadians – has received $2 million dollars as part of a national genomic contest led by Génome Canada.

Dr. Paul Goodyer’s team at the RI-MUHC is at the forefront of North American efforts to develop a novel drug for nephropathic cystinosis, a disorder that is 10 times more prevalent in Québec. Without treatment, all affected children require kidney transplantation and survival beyond the age of 30 is rare.

“This is crucial to taking the next steps toward setting up a clinical trial; current treatment is helpful but doesn’t prevent the eventual outcomes,” says Dr. Goodyer, who is a senior scientist from the Child Health and Human Development Program at the RI-MUHC and a pediatric nephrologist at the Montreal Children’s Hospital of the MUHC. “If successful, this new non-toxic drug will have a similar application in a wide variety of genetic diseases caused by Nonsense Mutations, including cancers, while providing significant cost-savings to the Canadian healthcare system.”

This form of nephropathic cystinosis is caused by Nonsense Mutations, an error in the gene code that prematurely signals the cell to stop producing a protein. In cystinosis, the loss of the CTNS protein causes a massive build-up of the amino acid cystine in all the cells of the

“At six months we realized something wasn’t right since Nolan was always thirsty and asking for water throughout the night but wasn’t gaining any weight or showing appetite. He could barely move or do anything that required physical demand. He refused everything. We brought him to the MUHC and straightaway they recognized the typical characteristics of the disease,” says Nolan’s mother Sophie, who has endured many sleepless nights tending to her son.
body, which causes gradual organ deterioration.

“Our drug overcomes Nonsense Mutations by tricking the cell into overlooking the mistake in the code and continuing protein production,” explains Dr. Goodyer, who is also a professor of Pediatrics in the Faculty of Medicine at McGill University. “By restoring production of the normal CTNS protein, sustained ELX-02 therapy has the potential to be curative.”

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Nolan, 9, has been in and out of hospitals since being diagnosed with the disease soon after birth.

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Dr. Goodyer has been collaborating with United States-based Eloxx Pharmaceuticals and expects clinical trials to eventually be concentrated in Montreal alongside Chicago and San Francisco. This project was one of seven to be awarded major funding from a nation-wide Genome Canada Genomic Applications Partnership Program (GAPP), which aims to foster partnerships between academic researchers and end users with projects that translate scientific innovation into action for the benefit of the Canadian population and economy.

“With this funding, local researchers will be capable of developing concrete and effective solutions that will save human lives, thanks to advances in genomics technology. Promising endeavors like Dr. Goodyer’s project are part of the shift towards precision medicine, which is transforming the way we treat rare diseases such as nephropathic cystinosis,” says Daniel Coderre, President and CEO of Génome Québec.

About the Research Institute of the MUHC

The Research Institute of the McGill University Health Centre (RI-MUHC) is a world-renowned biomedical and healthcare research centre. The Institute, which is affiliated with the Faculty of Medicine of McGill University, is the research arm of the McGill University Health Centre (MUHC) – an academic health centre located in Montreal, Canada, that has a mandate to focus on complex care within its community. The RI-MUHC supports over 420 researchers and close to 1,200 research trainees devoted to a broad spectrum of fundamental, clinical and health outcomes research at the Glen and the Montreal General Hospital sites of the MUHC. Its research facilities offer a dynamic multidisciplinary environment that fosters collaboration and leverages discovery aimed at improving the health of individual patients across their lifespan. The RI-MUHC is supported in part by the Fonds de recherche du Québec – Santé (FRQS). rimuhc.ca.

About Génome Québec

Génome Québec’s mission is to catalyze the development and excellence of genomics research and promote its integration and democratization. It is a pillar of the Québec bioeconomy and contributes to Québec’s influence and its social and sustainable development. The funds invested by Génome Québec are provided by the ministère de l’Économie, de la Science et de l’Innovation du Québec (MESI), the Government of Canada, through Genome Canada, and private partners. To learn more, visit genomequebec.com.

About the Funding

The Research Institute of the McGill University Health Centre received funds from Génome Québec, which are provided by the ministère de l’Économie, de la Science et de l’Innovation du Québec (MESI), the Government of Canada, through Genome Canada, and private partners. To know more about the research project: genomequebec.com/214-en/project/novel-aminoglycoside-readthrough-therapy-for-nonsense-mutations/

To know more about the research project: genomequebec.com/214-en/project/novel-aminoglycoside-readthrough-therapy-for-nonsense-mutations/
The North American Pediatric Renal Trials and Collaborative Studies and Horizon Pharma plc Announce New Long-Term Cystinosis Registry

Chicago - A prospective longitudinal natural history registry designed to provide long-term data evaluating people living with cystinosis was launched today at a meeting of The North American Pediatric Renal Trials and Collaborative Studies (NAPRTCS) in Chicago, Ill. The registry will collect and organize physician-reported data that will help healthcare professionals better understand the natural history of cystinosis. Data collected will also be available to researchers exploring new potential treatments for cystinosis. Utilizing the established NAPRTCS infrastructure which collects data on patients with chronic kidney disease, on dialysis and following kidney transplant, the cystinosis registry was made possible through a partnership between NAPRTCS, leading physicians in the cystinosis community, and the medical leadership at Horizon Pharma plc (Nasdaq: HZNP).

"Initial cystinosis natural history studies were conducted decades ago by the National Institutes of Health and there have been few studies since that time that track the progression of cystinosis," said Bradley Warady, M.D., professor of pediatrics at the University of Missouri-Kansas City School of Medicine, director of pediatric nephrology at Children’s Mercy Kansas City and member of the NAPRTCS board of directors.

"With recent advancements, people with cystinosis are living longer lives; however, our understanding of the disease in the context of these recent advancements and new treatments is limited. By gathering data from people living with cystinosis over an extended period of time, the NAPRTCS registry will provide key information for physicians and researchers."

People living with cystinosis under the age of 25 can be enrolled in the study, regardless of what therapies or medications they are receiving.

“This registry will help us better understand cystinosis, identify management improvements, and explore new treatment options that arise,” said Paul Grimm, M.D., professor of pediatrics (nephrology) at the Lucile Salter Packard Children’s Hospital at Stanford. “The registry will also help us answer a number of questions related to the long-term outcomes for patients and the long-term effects of cystinosis on the kidneys and all other organ systems.”

While Horizon provided NAPRTCS financial support to develop the registry, the company does not own any of the data – it belongs entirely to the medical community and is accessible through a securely protected online portal managed by NAPRTCS. Interested healthcare professionals should visit naprtcs.org.

“This registry is an example of a prioritized initiative based on input we’ve heard from physicians and researchers,” said Jeffrey Kent, M.D., senior vice president, medical affairs, Horizon Pharma plc. “A key component of understanding the opportunity for new medicines for rare diseases is understanding the natural history of the disease. In order to do that, there needs to be a system where this information can be collected. This registry will allow..."
the medical community to gather and synthesize data that can be evaluated to better understand cystinosis and explore new approaches that can advance the treatment of the disease.”

About NAPRTCS
The North American Pediatric Renal Trials and Collaborative Studies (NAPRTCS) is a research effort organized in 1987. The NAPRTCS patient registry follows the clinical course and natural history of children with renal dysfunction from participating sites across North America. It follows these patients as they move across the treatment continuum from chronic kidney disease to dialysis and transplantation. For more information, visit naprtcs.org.

About Horizon Pharma plc
Horizon Pharma plc is focused on researching, developing and commercializing innovative medicines that address unmet treatment needs for rare and rheumatic diseases. By fostering a growing pipeline of medicines in development and exploring all potential uses for currently marketed medicines, we strive to make a powerful difference for patients, their caregivers and physicians. For us, it's personal: by living up to our own potential, we are helping others live up to theirs. For more information, please visit horizonpharma.com, follow us @HZNPplc on Twitter, like us on Facebook or explore career opportunities on LinkedIn.

Forward-Looking Statements
This press release contains forward-looking statements, including statements regarding the potential benefits of the cystinosis registry. Forward-looking statements speak only as of the date of this press release and Horizon Pharma does not undertake any obligation to update or revise these statements, except as may be required by law. These forward-looking statements are based on management’s expectations and assumptions as of the date of this press release and actual results may differ materially from those in these forward-looking statements as a result of various factors. These factors include, but are not limited to, whether physicians and patients provide information to the registry and whether the registry is ultimately used in the medical community. For a further description of these and other risks, please see the risk factors described in Horizon Pharma’s filings with the United States Securities and Exchange Commission, including those factors discussed under the caption “Risk Factors” in those filings. Forward-looking statements speak only as of the date of this press release and Horizon Pharma undertakes no obligation to update or revise these statements, except as may be required by law.

We’d Love to Hear From You
We are always looking for the latest news from around the global cystinosis community. This includes your stories! To see your words in the next newsletter, send article ideas to our editor at adelmann.aimee@gmail.com.
Horizon Pharma is pleased to invite you to a program designed specifically for people living with cystinosis and their families. 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 nephropathic cystinosis
- Participate in discussions and exercises, including an informative presentation about PROCYSBI

Tentative meeting locations for 2018 and 2019 are Atlanta, Georgia; Portland, Oregon; Sacramento, California; Miami, Florida; Charlotte, North Carolina; Kansas City, Missouri; Dallas, Texas; Houston, Texas; Cleveland, Ohio; Virginia Beach, Virginia; Minneapolis, Minnesota; Chicago, Illinois; Boston, Massachusetts; Pittsburgh, Pennsylvania; Rochester, New York.

To RSVP to one of these cities or to request a program near you, please call 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.
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 vomiting, 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.
Hello CRN advocates!
I’m so excited to be the new Vice President of Education and Awareness. This is my first time putting a newsletter together and I hope you enjoy it! As you can see there is a new look and feel to this edition. A lot of thanks go to Christy, Clair, Clint, and the rest of the CRN board for helping to put together this beautiful, brand new newsletter!

As you can see from this edition, CRN has been very busy this year! We have been attending lots of events including American Society of Nephrology Convention, (see below), Global Genes Rare Patient Advocacy Summit, and having CRN meet-ups all over the county. This year our scholarship program was able to award four very deserving recipients (see page 3). I hope you find the articles as informative and impressive as I do. We have incredible and passionately active families and volunteers.

Our committee also has lots of ideas on how to broaden our outreach to educate and inform even more people about cystinosis. One of our goals is to help people understand that this is no longer a childhood disease. It’s a lifelong disease and we hope to help the community and professionals understand that.

For those of you who don’t know me I wanted to take a little time to introduce myself. I have been a patient and patient advocate for some time. I also serve on a variety of advisory boards to assure that organizations think about the patient perspective and incorporate those perspectives in their work. For my day job I work for Donate Life Northwest doing education and awareness about donation and transplant. I’m always happy to be a resource to anyone who has questions about transplant or is looking for resources. This fall I had a wonderful opportunity to travel to Croatia and Slovenia, as you can see from some of my pictures.

I am looking forward to a busy and productive year. My hope is that each of you will find a way to join us in actively promoting new and better understanding about cystinosis.

See you at the conference in July!
Happy Holidays!

By Aimee Adelmann, Vice President of Education and Awareness
CRN Visits American Society of Nephrology for Kidney Week 2018

By Aimee Adelmann, Vice President of Education and Awareness, Alison Moreno, and Carol Hughes

The American Society of Nephrology (ASN) KIDNEY WEEK 2018 was held in San Diego, California the last week in October. ASN leads the fight against kidney diseases by educating health professionals, sharing new knowledge, advancing research, and advocating the highest quality care for patients. This was a week-long event that included both pediatric and adult nephrologists in addition to researchers from around the world.

The Cystinosis Research Network has been attending the American Society of Nephrology convention for many years. It is always nice to run into familiar faces and meet many new ones as well. Carol Hughes, one of our CRN board members who attended the meeting, noted that one of the many highlights of the week was connecting with several of CRN’s Scientific Review and Medical Professional Advisory Boards; Drs. Rick Kaskel, Ewa Elenberg, Julie Inglefinger, Joshua Zaritsky and Craig Langman.

This year was especially unique because we had the opportunity to invite newer members of our community. To represent CRN we had; Aimee Adelmann, a cystinosis patient and CRN Vice-president of Education and Awareness; Carol Hughes, mom to Heidi and a long-time CRN Board Member; and Alison Moreno, wife of Ray Moreno, an adult living with cystinosis. All three of these women staffed the CRN booth. With a rich variety of backgrounds, they were able to inform and educate a multitude of people from the nephrology specialty, from physicians, to students and even many researchers. They also learned a great deal from the individuals that visited our booth. It was great to learn that cystinosis is becoming a more commonly known rare disease among providers and scientists which is thanks to all the work our community is doing to educate the public about cystinosis.

Alison shared “I’m so excited that CRN is able to participate in these events and provide a high level of education. CRN has played a large role in making this a known disease; these providers may not know the nitty gritty details of the disease but they have a basic understanding. It was a pleasure to represent the cystinosis community and we have created some new relationships at the conference that will only continue to benefit us.”
Global Genes Rare Patient Advocacy Summit

By Clinton Moore, President

It was very exciting to attend, for my first time ever, the Global Genes Rare Patient Advocacy Summit in Irvine, California this year. It was a pleasure to attend this along with two members of our community, Katie Morrison and Mika Covington.

On the first day, I attended sessions pertaining to nonprofits. Everything from how to properly manage, lead, do financial reports, create, motivate, and more were discussed. On the second day, I attended sessions pertaining more to patient and caregiver needs. Depression, caring for the caregivers, and advocating for yourself were some of the things discussed. There was an unbelievable amount of knowledge and experience that was shared at this meeting.

During these two days I also had opportunities to discuss various topics with industry partners and other nonprofits that were in attendance. Overall, I felt it was a great experience for myself, and I know Katie and Mika felt the same way. We are definitely looking forward to next year.

Save the Date: Rare Disease Day

Rare Disease Day is February 28, 2019. There's still time to get involved.

Contact Aimee Adelmann for details at adelmann.aimee@gmail.com.
Advocacy in Action: A Look Ahead

Our cystinosis advocates are preparing for an action-packed 2019. Here are a few of the events we are actively participating in. Cheers to a new year and new opportunities for cystinosis awareness!

The Pediatric Academic Societies (PAS) Meeting brings together thousands of pediatricians and other health care providers united by a common mission: improve the health and well-being of children worldwide. This international gathering includes researchers, academics, as well as clinical care providers and community practitioners. Presentations cover issues of interest to generalists as well as topics critical to a wide array of specialty and sub-specialty areas.

The Patient Congress is focused on incorporating the patient voice across the entire product lifecycle. Through three tailored tracks, examine the pieces integral to the patient journey of both small and large patient populations including patient access, adherence, and advocacy. With more than 250+ industry leaders representing life sciences companies, pharmacies, health plans, provider organizations, patient groups, and patient leaders, join the conversation around innovative and collaborative ways we can improve access and health outcomes for patients. Develop your organization’s patient-centric strategy from drug development through commercialization, break down internal silos, and improve patients’ lives across the entire continuum of care.

The American Transplant Congress is the Joint Annual Meeting of the American Society of Transplant Surgeons (ASTS) and the American Society of Transplantation (AST). ATC provides a forum for exchange of new scientific and clinical information relevant to solid organ and tissue transplantation and brings together transplant physicians, scientists, nurses, organ procurement personnel, pharmacists, allied health professionals and other transplant professionals. The educational offerings provide attendees the opportunity to learn cutting-edge advances in research and exchange of ideas and practice in the field of solid organ and tissue transplantation.
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**
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.

**CRN 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.