

# THE CYSTINOSIS Advocate

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## 2019 CRN Conference: July 18-20 in Philadelphia

The 2019 Family Conference will be held July 18-20 at the Hilton Philadelphia at Penn's Landing. The Hilton Philadelphia at Penn's Landing is 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 + Kitchen. Walk to Philadelphia attractions including the Liberty Bell, Independence Hall, Museum of the American Revolution and Blue Cross Riverink Summerfest.

For more information on the hotel please visit their website:

<http://bit.ly/2lqjFGZ>

Online Participant Registration  
<https://www.cystinosis.org/2019-crn-conference-participant-form/>

Download Participant Registration  
<http://bit.ly/2N1AFaJ>

Hotel  
<http://bit.ly/2KYFkru>

We look forward to seeing you in Philadelphia in July!

### Join us at the 2019 CRN Conference to:

- Learn about new research findings
- Receive updates from international cystinosis organizations
- Meet and renew friendships with other individuals and 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

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## The President's Letter

Every newsletter when I begin writing this letter I feel as though I am repeating myself. I usually talk about how busy the CRN has been over the past 6 months and what we have accomplished, and to be honest, this one isn't going to be any different. The CRN is extremely busy as it has been since I took this role. And now more than ever with the family conference only about 2 months away. The hard work and dedication from every member of this board is amazing. I'm forever grateful that they take time out of their lives and contribute to this fascinating organization. I recently took a look at the data from just our social media outlets alone and it's amazing to see the growth over the past year. It's clear to see we are reaching the community like never before. Community is what the CRN stands for. It's what we believe in. It's what we work so hard day after day for. A community that we ourselves also belong to.

There are a thousand things that can arise when dealing with this disease, all of which we are equipped to help you deal with. Whether it be a resource that we already have available or creating a connection with someone that can assist you better. If we don't have the answer, we will find it. Utilize us. It's what we are here for. The CRN is your advocacy group.

President of your advocacy group,  
Clinton Moore



*Chandler and Clinton Moore.*



*The Moore Family.*

# CRN 2019 Conference Draft Agenda

## Wednesday, July 17th

**3:00 pm – 8:00 pm**                      **CRN Board of Directors Meeting**  
Columbus Ballroom A                      Closed Session

## Thursday, July 18th

**12:00 pm – 5:00 pm**                      **Registration/Information Desk**  
Grand Ballroom Foyer

**5:00 pm – 7:00 pm**                      **CRN Welcome Reception**  
Grand Ballroom                              Hosts: CRN Board of Directors  
Complimentary

**7:00 pm – 10:00 pm**                      **Family Introductions/Networking**  
Grand Ballroom                              Hosts: Tim Wyman and Jen Wyman, VP Family Support

A great opportunity to put faces to names of each family attending the conference. Bring your entire family as this session will include introductions and reveal the third book in the “Journeys with Cystinosis” comic series. Author and illustrator, Kevin McCalla will be available to autograph complimentary copies of the book. In addition, families will all participate in a special project at the end of the evening!

## Friday, July 19th

**7:00 am – 8:30 am**                      **Breakfast**  
Grand Ballroom Foyer                      Complimentary

**8:00 am – 5:00 pm**                      **Registration/Information Desk**  
Grand Ballroom Foyer

**8:00 am – 5:30 pm**                      **Childcare Open**  
Columbus Ballroom

The CRN is proud to offer a day care for all the children and siblings of the conference. We will have a full staff of Nannies from the Philadelphia Nanny Company to watch the children. Childcare will be provided both Friday and Saturday from 8am-5pm, excluding lunch time. There will be age appropriate activities, games, and tons of crafts to make and take! Wii system and movies are also available for fun. We will have amazing entertainment coming in both mornings from 9-12!

Parents MUST sign their child into daycare each morning and SIGN OUT every time they come to get their child(ren). Parents need to take their children out of childcare for lunch. Dosing of medications can only be administered by parents-not nannies or volunteers. Snacks and plenty of water will be provided. Guaranteed to allow the kids to not only bond, but to have the time of their lives!

9:00 am - 12:00 pm: Party Princess and Superhero Performance! For all ages!

10:00 am - 1:00 pm: Photo Booth with Superheroes and Princesses! Mom and dad can join before or after lunch to get some great photos with the kids!

### **Childcare Open, continued**

3:00 pm – 5:00 pm: Sing me a Story Foundation

The kids will write & illustrate stories about whatever they want in their very own blank storybooks. Each story goes on to be transformed into a personalized song by artists & bands across the country, raising awareness about cystinosis through the power of music.

[www.singmeastory.org](http://www.singmeastory.org)

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**8:15 am – 8:30 am**

Grand Ballroom

### **Welcome and Opening Remarks**

Clinton Moore, President

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**8:30 am – 9:30 am**

Grand Ballroom

### **Keynote Speaker**

David Fajgenbaum, MD, MBA, MSc

Dr. David Fajgenbaum, MD, MBA, MSc, FCPP, is the co-founder and Executive Director of the Castleman Disease Collaborative Network (CDCN), Assistant Professor of Medicine in Translational Medicine & Human Genetics at the University of Pennsylvania, and Associate Director, Patient Impact for the Penn Orphan Disease Center. Dr. Fajgenbaum is also a patient battling idiopathic multicentric Castleman disease (iMCD). As common as ALS and as deadly as lymphoma, iMCD involves the immune system attacking and shutting down the body's vital organs (liver, kidneys, bone marrow, heart). He became ill during his third year of medical school in 2010, spent five months hospitalized, had his last rites read, and has had four deadly relapses since. In 2012, Dr. Fajgenbaum began conducting Castleman disease research at Penn and co-founded the CDCN, through which he has spearheaded the "Collaborative Network Approach." He currently leads the Castleman Research Program at Penn as PI of 18 translational research studies, including an international natural history study and the first-ever NIH R01 grant studying iMCD. As a result of his work, Fajgenbaum is currently in his longest remission ever thanks to a treatment that he identified, which had never been used for iMCD.

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**9:30 am – 9:50 am**

Grand Ballroom

### **CRN – Your Advocacy Group**

Christy Greeley, Executive Director, VP Research

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**9:50 am – 10:10 am**

Grand Ballroom

### **Cystinosis Network Europe Update**

Anne Marie O'Dowd, Chair, Cystinosis Network Europe

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**10:10 am – 10:30 am**

Grand Ballroom

### **The Promise of New Drug Targets to Reverse Renal Injury in Cystinosis**

Supported by HRB Ireland and Cystinosis Foundation Ireland  
Minnie Sarwal, MD, FRCP, DCH, PhD

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**10:30 am – 10:45 am**

Grand Ballroom Foyer

### **Break**

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**10:45 am – 11:10 am**

Grand Ballroom

### **Cystinosis – A Review of Old and New**

Joshua Zaritsky, M.D.

A review of the epidemiology, pathophysiology and treatment options of cystinosis.

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**11:10 am – 11:35 am**

Grand Ballroom

### **Anticipating Renal Replacement Therapy**

Anticipating Renal Replacement Therapy

Paul Grimm, MD, Stanford

### **Anticipating Renal Replacement Therapy, continued**

Unfortunately, even when children are diagnosed with cystinosis at a young age and they are able to tolerate the cystine depleting therapy, there may be substantial damage already done to the kidneys. This damage may occur without the usual tests of kidney function being abnormal. The creatinine might be “normal” even after significant amounts of kidney damage has occurred. We are usually born with a lot of extra kidney function, so we don’t start feeling ill from kidney failure until we are down at 10 or 15% function. Therefore we have to rely on the kidney numbers that come with routine blood testing, but they are not so easily interpreted. So, as a parent or a patient, how worried should you be about your kidney numbers? What are the kidney numbers that are good, or bad, or worse? Are there any special considerations for a cystinosis patient compared to any other patient with Chronic Kidney Disease?

We will discuss the kidney numbers that include CKD Stages (1, 2, 3, 4 and 5/ESRD) serum creatinine, BUN and a relative newcomer called Cystatin C. These are used singly and in combination to get a better understanding of the kidney function. We may also discuss measuring protein loss in the urine as a way to help determine how far advanced the kidney function is.

We will talk about plotting these kidney numbers on a graph and using it to help predict when the kidney function might be falling to the level where some kind of kidney replacement therapy/renal replacement therapy will be necessary. At the end of the session, we will also discuss ways to slow the progress of the kidney failure; including nutritional choices, lifestyle choices and medication.

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**11:35 am – 12:00 pm**  
Grand Ballroom

**A Systematic Review of Adult Complications of Cystinosis**  
Rachel Kasimer, Medical Student/Craig Langman, MD

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**12:00 pm – 1:00 pm**  
Grand Ballroom Foyer

**Lunch**  
Complimentary

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### **Panel Sessions 1, 2, & 3**

Panel Sessions are targeted towards different stages of the cystinosis journey, divided into tracks by age. They will be smaller group sessions with a panel of experts presenting information, guiding discussion and fielding questions. Attendees should feel free to attend sessions of most interest to them. Some sessions are closed as noted below.

**1:00 pm – 2:00 pm**  
Grand Ballroom A

**Panel Session 1**  
**Caregiver/Infant & Child (0-10): Cystinosis 101**

Panelists: Drs. Katharina Hohenfellner, Larry Greenbaum, Joshua Zaritsky, Mihir Thacker, Ranjan Dohil, Jess Thoene, Ewa Elenberg, and Paul Goodyer

Grand Ballroom B/C

**Teenager (11-17): Cystinosis 201**

Panelists: Drs. Rachel Bishop, Craig Langman, Rick Kaskel, Doris Trauner, Minnie Sarwal, Neveen Soliman, Invited panelists

Grand Ballroom D

**Adult (18+): Cystinosis 301**

Panelists: Drs. William Gahl, Galina Nesterova, Paul Grimm, Maya Doyle, Patrick Gipson

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<p><b>2:00 pm – 3:00 pm</b> Grand Ballroom A</p>	<p><b>Panel Session 2</b>  <b>Caregiver/Infant &amp; Child (0-10): Physician Q &amp; A</b>            Panelists: Drs. Katharina Hohenfellner, Larry Greenbaum, Craig Langman, Paul Grimm, Joshua Zaritsky, Ranjan Dohil, Jess Thoene, Ewa Elenberg, Neveen Soliman, Paul Goodyer, Mihir Thacker, Invited Panelists</p>
<p>Grand Ballroom B/C</p>	<p><b>Teenager (11-17): Social Relationships and Gaining Independence</b>            (Session for teens 11-17 and their parents)            Panelists: Maya Doyle, MSW, PhD, LCSW, Carrie Ostrea, Seth Rotberg, Ina Gardener, MEd, LPC</p>
<p>Grand Ballroom D</p>	<p><b>Adult (18+): Living with Cystinosis as an Adult: Healthcare, Expectations, Self-Management, Reproductive Issues</b>            (Closed session for adults 18 + and their partners)            Panelists: Drs. Rachel Bishop, William Gahl, Galina Nesterova, Patrick Gipson, Elena Levchenko (via Skype)</p>
<p>Room TBD</p>	<p><b>Adult (18+): Living with Cystinosis as an Adult: Healthcare, Expectations, Self-Management, Reproductive Issues</b>            (Closed session for parents of adults living with Cystinosis)            Panelists: Colleen Hammond, Carol Hughes            The purpose of the session is intended to be a private setting for parents of adults with cystinosis to share information, insight, advice and encouragement. This session will provide an opportunity to learn from others who face similar challenges, and allow you to talk about your experiences.</p>
<p><b>3:00 pm – 4:00 pm</b> Grand Ballroom A</p>	<p><b>Panel Session 3</b>  <b>Caregiver/Infant &amp; Child (0-10): Neurocognitive &amp; Educational Issues</b>            Panelists: Marybeth Krummenacker, Doris Trauner, MD, Neveen Soliman, MD</p>
<p>Grand Ballroom B/C</p>	<p><b>Teenager (11-17): Transplant and Dialysis</b>            Panelists: Drs. Rick Kaskel, Paul Grimm, Minnie Sarwal, Patrick Gipson</p>
<p>Grand Ballroom D</p>	<p><b>Adult (18+): Adults Living with Cystinosis: Focus on Mental Health and Wellness</b>            (Closed session for Cystinosis adults and their partners)            Panelists: Maya Doyle, MSW, PhD, LCSW and Carrie Ostrea, Ina Gardener, MEd, LPC, Seth Rotberg, Galina Nesterova, MD</p>
<p><b>4:00 pm – 4:15 pm</b> Grand Ballroom Foyer</p>	<p><b>Break</b></p>
<p><b>4:15 pm – 5:30 pm</b> Grand Ballroom</p>	<p><b>Medical Panel</b>            Moderator: William A. Gahl, M.D., Ph.D.            Please join the entire group for the unique and informative opportunity to have your questions and concerns addressed by the leading physicians and researchers in cystinosis. All of the doctors who have presented at the Family Conference, all attending Medical Advisory Board and Scientific Review Board members, as well as other health care professionals involved in treating and researching cystinosis are scheduled to participate. Questions for the panel will be collected during the proceedings today.</p>

**5:30 pm**

Grand Ballroom

**Group Photograph**

All conference attendees should gather for a group photograph and a photograph of all individuals in attendance living with cystinosis. Please bring your green conference t-shirt to wear for a very special group photo! Also, please pick your children after the Medical Panel from Childcare and bring to the Grand Ballroom for this wonderful remembrance of the conference.

**6:30 pm – 9:30 pm**

Grand Ballroom

**Meet & Greet Family Buffet Dinner**

Hosts: Adult Leadership Advisory Board (ALAB)

Complimentary

Take time to refuel and relax with a casual dinner complete with good company. Share your learnings from the day and catch up with other families during our meet and greet buffet dinner.

**7:00 pm – 10:00 pm**

R2L Restaurant

50 S. 16th Street

**Speaker/VIP Dinner**

Hosts: CRN Board of Directors

Closed Session

**Saturday, July 20th**

**7:00 am – 8:30 am**

Grand Ballroom Foyer

**Breakfast**

Complimentary

**8:00 am – 4:00 pm**

Grand Ballroom Foyer

**Registration/Information**

**8:00 am – 5:00 pm**

Columbus Ballroom

**Childcare Open**

9:00 am-12:00 pm: Fabulous Faces of Philly - Fabulous Balloon Twisting, Balloon twisting school, Face painting and tattoos! For all Ages!

1:00 pm-5:00 pm: Philadelphia Nanny Company to supervise in room activities!

Tween/Teen- Field Trip and Team Building to Escape the Room Philly! (Must have permission slip signed, for ages 12-19)

**8:30 am – 8:45 am**

Grand Ballroom

**Opening Comments**

Christy Greeley, Executive Director and VP Research

**8:45 am – 10:15 am**

Grand Ballroom

**Advocacy**

**8:45 am – 9:15 am**

Grand Ballroom

**Carrie Ostrea**

This interactive session will provide guidance and education on how to be a stronger advocate for the cystinosis community. This session will explain the various types of advocacy to help each participant determine what they are most passionate about and what their next steps should be to pursue that direction. In addition, this will focus on tools and strategies to empower patients and their families to share their story impactfully with your community, local media, your healthcare team, and on social media.

<b>9:15 am – 9:45 am</b> Grand Ballroom	<b>Seth Rotberg</b> This session will go through the patient journey of Seth Rotberg -a young adult in the rare disease space- and how he was able to turn challenges into opportunities. It will discuss how a person impacted by a rare disease can use his or her diagnosis as motivation to get more involved in fundraising, advocacy, and volunteer efforts. This session will also discuss some of the different ways to advocate for yourself and the next steps to take for a better tomorrow. <a href="https://www.youtube.com/watch?v=5_O5TfMVqD8">https://www.youtube.com/watch?v=5_O5TfMVqD8</a> <a href="https://www.youtube.com/watch?v=84QwwHOizGE">https://www.youtube.com/watch?v=84QwwHOizGE</a>
<b>9:45 am – 10:30 am</b> Innovation A & B	<b>Breakout Session</b> Teens and adults living with cystinosis to meet with Adult Leadership Advisory Board (ALAB) members to discuss ideas and resources for the teen and adult community.
<b>9:45 am – 10:15 am</b> Grand Ballroom	<b>Jean Campbell</b> (communiation/networking)
<b>10:15 am – 10:30 am</b> Grand Ballroom Foyer	<b>Break</b>
<b>10:30 am – 1:00 pm</b> Grand Ballroom	<b>Research Updates</b> Host: Paul Grimm, MD Clinical research updates from around the world. Brief updates will be shared from a variety of researchers in order to better understand the future of cystinosis diagnosis and management.
<b>10:30 am – 10:50 am</b>	<b>Stem Cell Gene Therapy for Cystinosis: description of the upcoming clinical trial</b> Stephanie Cherqui, PhD
<b>10:55 am – 11:15 am</b>	<b>A No-Nonsense Approach to Cystinosis</b> Paul Goodyer, MD
<b>11:20 am – 11: 40 am</b>	<b>Muscle Weakness in Patients with Cystinosis</b> Larry Greenbaum, MD, PhD, FAAP Muscle weakness is a complication of cystinosis. It tends to worsen over time. It is most commonly seen in adults with cystinosis. Many believe that cysteamine therapy prevents or delays the development of muscle weakness. Grip strength is one way of measuring muscle strength. We have used grip strength to measure muscle strength in children in the CKiD study. This is a study sponsored by the NIH of approximately 1000 children with chronic kidney disease. It only includes a small number of children with cystinosis. We showed that children with chronic kidney disease have decreased grip strength compared to healthy children. At our clinic at Emory, we have conducted a study of grip strength of children and adults with cystinosis. In order to study more patients, we measured grip strength of children and adults with cystinosis at the last CRN Conference in Utah. We were very appreciative of the many children and adults who participated in the study. We also asked patients about problems with weakness in their daily lives and if they have had times during their lives when they did not take cysteamine. In this study, we showed

### **Muscle Weakness in Patients with Cystinosis, continued**

that grip strength was decreased in patients with cystinosis. We are now measuring grip strength every 1-2 years in patients with cystinosis to see if it decreases over time.

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**11:45 am – 12:05 pm**

### **Genetic Newborn Screening for Cystinosis and SMA in Germany**

Katharina Hohenfellner, MD

Newborn screening (NBS) programs for treatable metabolic disorders have been enormously successful, but molecular-based screening has not been broadly implemented so far. This prospective pilot study was performed within the German NBS framework. DNA, extracted from dried blood cards collected as part of the regular NBS program, was screened for cystinosis and spinal muscular atrophy (SMA). Between January 15, 2018 and May 30 2019, more than 260, 000 newborns were screened in Germany for cystinosis and SMA. Until now 1 patient with cystinosis and 26 patients with SMA have been diagnosed. No false screening result have been reported so far. Communication of findings to parents, and confirmation of diagnosis were accomplished in a multi-disciplinary setting and the program was well accepted by hospitals, physicians, and parents. This pilot study demonstrates the efficacy of a molecular-based neonatal screening program for cystinosis and SMA using an existing national screening framework.

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**12:10 pm – 12:30 pm**

### **The Lifecourse Journey of Cystinosis**

Frederick Kaskel, MD, PhD, FAAP, FASN

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**12:35 pm – 12:55 pm**

### **Foxe Labs CRN Neurocognitive Research Update**

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**1:00 pm – 2:00 pm**

Grand Ballroom Foyer

### **Lunch**

Complimentary

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**2:00 pm – 3:00 pm**

Columbus Ballroom Foyer

### **Poster Session**

This session will showcase a mix of science, medicine, industry, advocacy group and patient experiences to provide an interactive experience for both family and professional attendees. Researchers, clinicians, industry, advocacy representatives, students, patients, and caregivers will be invited to exhibit their latest research findings, treatment breakthroughs, advocacy group updates and real patient and family experiences. This will be an interactive session where exhibitors will be available to discuss their work or experiences with those attending. We invite you to browse the posters and take this opportunity to ask the authors and presenters questions.

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**3:00 pm – 4:00 pm**

Grand Ballroom

### **Parents of Children and Adults with Cystinosis Panel**

Moderator: Jen Wyman, CRN VP Family Support

Panelists: Invited

Panel presentation during which parents of children and adults with cystinosis will answer prepared questions and address topics related to the use of coping mechanisms through the ups and downs that cystinosis brings related to not only developmental and transitional issues of daily life but also medical issues. Parents of individuals at every stage of the disease will be featured and will share how they have managed the variety of challenges they have faced. Audience participation will be encouraged.

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**4:00 pm – 5:00 pm**

Grand Ballroom

### **Adults Living with Cystinosis Panel**

Moderator: Carrie Ostrea

Panelists: Adult Leadership Advisory Board (ALAB) members, Invited panelists

Panel presentation during which individuals living with cystinosis will answer prepared questions and address topics related to the use of coping mechanisms and strategies for success through the ups and downs that cystinosis brings. Audience participation will be encouraged. Adult Leadership Advisory Board members in attendance will participate.

ALAB is a new CRN initiative and is comprised of adults living with cystinosis. Ultimately, this program will develop programming and opportunity for adults affected by cystinosis.

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**5:00 pm – 5:15 pm**

Grand Ballroom

### **Closing Remarks**

Clinton Moore, CRN President, Christy Greeley, Executive Director

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**6:00 pm – 10:00 pm**

Grand Ballroom

### **Farewell Dinner Dance**

Complimentary

All conference attendees – pack your dancing shoes for our final event of the week – the dinner dance! Before saying goodbye, recharge with a delicious dinner then show your moves on the dance floor! After all, this evening is to celebrate YOU and our cystinosis community...Ring in the Future!

## **CRN 2019 Conference Poster Session Requirements**

- The Poster Session will take place during the conference proceedings on Saturday, July 20. This session will showcase a mix of science, medicine, industry and advocacy group, and patient experiences to provide an interactive experience for both family and professional attendees. Researchers, clinicians, industry, advocacy representatives, students, patients, and caregivers will be invited to exhibit their latest research findings, treatment breakthroughs, advocacy group updates, and real patient and family experiences. This will be an interactive session where exhibitors will be available to discuss their work or experiences with those attending. Attendees will be invited to browse the posters and take this opportunity to ask the authors and presenters questions.
  - Posters may be submitted into one of the following three categories:
    - Research/Clinical, Industry/ Advocacy, or Patient/Family.
    - Research Poster Presenters are asked to provide two hand-outs describing their work: one is to be a brief discussion of the exhibit written in layman's terms; the second, a more detailed, technical discussion for the attending professionals. These hand-outs are to be available at the poster exhibit for the benefit of all those attending this session.
    - Abstracts are to be provided for enclosure in the conference notebook. Abstracts should specify to which of the three categories they apply and should include a brief description of the information to be included on the poster. Please also include the main contact name, email address, daytime telephone number, title, and affiliation, if applicable.
    - Poster session abstracts may be submitted to Christy Greeley
- via email (as .doc or .pdf files) to [cgreeley@cystinosis.org](mailto:cgreeley@cystinosis.org) no later than June 15, 2019. Questions may be submitted via email or by telephone at (847) 204-6004.
- Each poster should have at least one presenter who must remain with their poster throughout the session.
  - Posters should be no larger than 4x4', and will be displayed on bulletin boards. Please specify if you require other display methods.
  - All costs associated with creation of the poster will be the responsibility of the entrants.
  - Entrants must give permission for poster abstracts to be published with conference materials, as well as posted on the CRN website ([www.cystinosis.org](http://www.cystinosis.org)).
  - Space is limited, and the CRN Conference Planning Committee reserves the right to reject any submitted abstract.



## Breaking Barriers Together: New Cystinosis Adult Leadership Group



Take a look at these faces.

A few decades ago, their parents were told they had cystinosis. They were also told, without proper treatment, they might not live to be teenagers.

Fast forward to 2019. Research is advancing. For most, medication options exist. Need a kidney transplant? Donor chains and online campaigns have helped pair us with organ donors who were once complete strangers. Talking to someone with this rare disease used to require travel. Now we have the internet and social media to connect

us almost instantaneously.

Opportunities have grown for our adults with cystinosis and now we face a new sets of challenges. In our continued commitment to an evolving cystinosis community, the CRN has formed a new Adult Leadership Advisory Board (ALAB). Each ALAB member is an adult living with cystinosis and is participating in a training program to foster success. Through ALAB, members will develop programming to inform, include and advance adults affected by cystinosis. They will partner with CRN board

members and other organizations to address the issues and provide mentorship.

Our goal is to create a group that will not only benefit from the experience, but will contribute to both the ALAB program and the entire cystinosis community.

There is a saying, "Alone we go faster, together we go further." We do not have to walk this path alone. Let's go further together!

Get in touch with an ALAB member or view their profiles at [cystinosis.org](http://cystinosis.org) under the Support & Resources tab.



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dispensing pharmacy for Cystaran.*

#### *\*Storage Information*

**When unopened:** Store in the freezer, in the original carton.

**When opened:** Keep between 36°-77°F (2°-25°C)

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

**Discard bottle:** After 7 days

**At school:** Discuss with your local school the options for your child, as rules for each school district vary.

#### *Order Information*

Please call: **AllianceRx Walgreens Prime Specialty Pharmacy**

**at 1-877-534-9627** Monday-Friday 8am to 7pm EST and  
Saturday 9am to 5pm EST

*You are encouraged to report negative side effects of prescription drugs to the FDA.  
Visit [www.fda.gov/medwatch](http://www.fda.gov/medwatch) or call 1-800-FDA-1088*

  
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## Advocacy in Action: A Look Ahead



CRN will join the American Society for Nephrology (ASN) and more than 13,000 other kidney professionals from across the globe at **Kidney Week 2019** in Washington, DC. The world's premier nephrology meeting, Kidney Week provides participants exciting and challenging opportunities to exchange knowledge, learn the latest scientific and medical advances, and listen to engaging and provocative discussions with leading experts in the field.



As the largest gathering of rare disease patients, caregivers, thought leaders and other rare disease stakeholders in the world, the **RARE Patient Advocacy Summit** is an unparalleled opportunity to forge meaningful connections with other rare advocates and take home actionable strategies and tools to accelerate change. This year, the Summit will take place in San Diego at the Sheraton San Diego Hotel & Marina (<https://globalgenes.org/event/patient-summit/>). CRN will be in attendance once again this year at this important gathering of advocates.

## CRN Announces 2019 Academic Scholarship Deadline



CRN is currently accepting academic scholarship applications which are available to individuals and siblings of individuals with cystinosis. Cystinosis can present a financial burden on families; therefore these awards have been made available to provide supplemental financial assistance to a student. The \$1000 awards are

awarded yearly to an individual who is accepted into a post-secondary school.

The CRN Scholarship Award and the Sierra Woodward Sibling Scholarship Award applications deadline is August 1, 2019. Information and applications can be downloaded from the CRN website at [cystinosis.org](http://cystinosis.org) under Support & Resources.

*2018 Scholarship winners (top left to bottom right): Kathleen Roberts, Jack Kitchens, Rhyana Christine Juutilainen, and Emily Ann Bogan.*

## Deanna Lynn Potts Scholarship



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:**

1. Documentation/verification of cystinosis (e.g. letter from physician.)
2. An official copy of high school transcript.
3. Two letters of recommendation from current teachers/faculty members and/or counselors regarding applicant's scholastic

aptitude and personal qualifications.

4. An essay of 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.

**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 1st of each year. You can call the Cystinosis Foundation for an application 888-631-1588 or go to [www.cystinosisfoundation.org](http://www.cystinosisfoundation.org). Print the Application and mail it to the Cystinosis Foundation.

# Cystinosis Ireland Research Funding Opportunities 2019

Please note that both of these research funding schemes support researchers and research activity based outside of Ireland.

The Cystinosis Ireland Seedcorn Funding scheme aims to provide researchers with the opportunity to generate solid preliminary data which would contribute to a larger, sustainable, longer-term application for funding.

Researchers that are new to the field of cystinosis are particularly welcome. We also welcome applications from researchers from scientific fields other than Biosciences and Clinical Sciences where there the potential impact or application of the proposed research is relevant to patients and families living with cystinosis. This includes scientists in social sciences, engineering and other disciplines.

We are particularly open to new, high-risk, blue-sky applications and will consider these favourably (Applicants should highlight this in their application).

In particular, this seedcorn fund may be used to develop highly competitive research proposals that are suitable for submitting to a future MRCG-HRB co-funding call (see below: open for applications in September 2019)

Although typical projects funded tend to be in the region of €10,000, the Cystinosis Executive will consider applications that are for more than this (even substantially more) where the project is well justified and is relevant to patients and families living with cystinosis. All research proposals are subject to peer review.

**MAXIMUM FUNDING AVAILABLE:**  
€10,000 per project (more where justified)

**DURATION OF PROJECTS:** 2-6 months (longer where justified)

**CLOSING DATE FOR**

**APPLICATIONS:** 2 August 2019; 29 November 2018.

Please note that this funding scheme supports researchers and research activity based outside of Ireland.

Researchers interested in this scheme should email [research@cystinosis.ie](mailto:research@cystinosis.ie) for an application form and to discuss any potential applications.

## **MRCG-HRB CO-FUNDING SCHEME 2019**

Cystinosis Ireland is very interested in supporting high quality research proposals to the MRCG HRB co-funding scheme. The next call for proposals is expected to open in September 2019.

Cystinosis Ireland has a strong track record of supporting research proposals for funding through this co-funding scheme and currently has two active projects underway. MRCG HRB awards are typically in the region of €100,000 per annum for up to 3 years.

Cystinosis Ireland encourages researchers who might consider applying to this scheme to also look at the Cystinosis Ireland seedcorn fund as a mechanism to generate preliminary results and to improve the competitiveness of a potential proposal to the MRCG-HRB co-fund scheme.

Please note that this funding scheme supports researchers and research activity based outside of Ireland.

We encourage researchers to contact us at [research@cystinosis.ie](mailto:research@cystinosis.ie) at any time in order to discuss your research ideas to see if it might be suitable for this funding scheme.

## **BACKGROUND TO CYSTINOSIS AND CYSTINOSIS IRELAND**

Cystinosis is a rare genetic disease that causes the amino acid cystine to accumulate in the body due to mutations in the CTNS gene. Cystinosis is classified as an 'orphan disease' by EURODIS – Rare Diseases Europe.

It is estimated that there are approximately 2,000 people worldwide diagnosed with cystinosis. As of May 2017, there are a total of 21 confirmed diagnoses of cystinosis in Ireland.

The severest form, infantile nephropathic cystinosis, causes kidney failure before the age of 10. Treatment is currently limited to cysteamine a cystine depleting drug which slows but does not cure the progression of the disease. All of those diagnosed in Ireland have infantile nephropathic cystinosis. Cystinosis Ireland was founded in 2003 as an Irish registered charity. It was created by volunteers – family members and family friends of those living with cystinosis. Its purpose is dedicated to raising money to fund research into cystinosis in Ireland and all over the world.

Cystinosis Ireland works closely with Temple Street Children's Hospital and Beaumont Hospital in Dublin as well as with the Great Ormond Street Hospital in London. Cystinosis Ireland also maintains partnerships with other cystinosis charities abroad including; The Cystinosis Foundation UK, the Cystinosis Research Foundation (CRF) in Irvine, California, USA, the Cystinosis Research Network (CRN) in Lake Forest, Illinois, US and the Canadian foundation – Cystinosis Awareness Research Effort (CARE). Through these partnerships, we share research findings, discuss drug access programmes, review challenges being faced by the greater community and work towards finding a cure. Cystinosis Ireland is also an active member of the Cystinosis

Network Europe and EURORDIS, the European rare disease patient group alliance.

## RESEARCH AREAS OF INTEREST

Since its foundation, Cystinosis Ireland has supported research projects to the value of €1.9 million in areas of cystinosis research aimed at better understanding this disease, developing better treatment options and ultimately seeking a cure. In this context, Cystinosis Ireland has worked with the Irish Health Research Board (through the MRCG-HRB co-funding scheme) and it has also developed its own seedcorn funding scheme (see below for more information).

Examples of research projects that Cystinosis Ireland has supported include:

- Drug eluting contact lenses for cystinosis therapy.
- Developing human stem cell models for cystinosis and therapeutic potential of aspartate.
- Unravelling the mechanisms of azoospermia and potential future treatments in Male cystinosis treatments.
- Targeting autophagy in nephropathic cystinosis.

In addition to these areas, Cystinosis Ireland is also particularly interested in the following research topics:

Side effects of cysteamine – halitosis and body odour become major issues as children age and this interferes with the level of adherence to cysteamine. It also causes major psychological issues. Work had been done on developing new compounds but something that would work with cysteamine would be very welcome. Concern about other side effects of cysteamine – for example, Ehler Danlos-type symptoms, collagen issues and subsequent death in one patient. What other effects is it having that we don't know about?

Muscle weakness – swallowing

problems have led to aspiration and death in some patients plus weakness in other muscles such as hands, legs, arms. Even patients who adhere well to their drug therapy have these issues. Is it caused by cysteamine and/or the cystinosis disease?

Bone Issues, including knock knees, which occur in most children whatever level of drug therapy adherence; spontaneous fractures and weak bones. Nearly all children have flat feet/fallen arches. These are in children who are well maintained on vitamin D, calcium etc and who have had diagnosis at birth.

In early childhood, not eating is a major issue. Most children need a g-tube for feeding after diagnosis. There's a question as to whether the cysteamine treatment of the cystinosis itself is the main reason for lack of appetite/not eating. A lack of eating has knock-on effects for life.

Developing models of care transition from childhood care to adult care in the health service.

Examining the social impacts of long-term childhood disease and also the impacts of treatment(s) on patient lives.

In addition to the above, Cystinosis Ireland strongly encourages researchers that are new to cystinosis to contact us.

We welcome applications from researchers from scientific fields other than Biosciences and Clinical Sciences where there the potential impact or application of the proposed research is relevant to patients and families living with cystinosis. This includes scientists in social sciences, engineering and other disciplines.

**We are particularly open to new, high-risk, blue-sky ideas from any discipline that might have**

**a positive impact on the lives of cystinosis patients either now or in the future.**

## NEW RESEARCHERS

Cystinosis Ireland strongly encourages researchers that are new to cystinosis to contact us. We also welcome applications from researchers from scientific fields other than Biosciences and Clinical Sciences where there the potential impact or application of the proposed research is relevant to patients and families living with cystinosis. This includes scientists in social sciences, engineering and other disciplines. Research | Cystinosis Ireland Cystinosis Ireland is one of the main patient organisations in Europe driving research into this rare disease. Since its establishment in 2003, Cystinosis Ireland has supported research projects focused on all aspects of this disease to the value of €1.9 million either through direct funding or as a co-funding partner with Ireland's national health research funding agency, the Health Research Board. Registered Office: 1-2 Cavendish Row, Dublin 1, Ireland. Charities Regulatory Authority No: 20053796

## Ireland Update: The Annual Cystinosis Ireland Family Day and the 5th Annual Dublin Cystinosis Workshop 2019

Cystinosis Ireland recently hosted a very successful annual Cystinosis Ireland Family Day in conjunction with the 5th Annual Dublin Cystinosis Workshop 2019.

This year the events, which ran over two consecutive days on 26 and 27 April, combined a ½ day of family-focused event with a 1½ day scientific workshop aimed at the scientists, clinicians and healthcare professionals.

Eleven families took part during the family event which included a number of practical talks and a question and answer session between the families and a panel of eight clinicians and healthcare specialists. The children also had their own programme of activities where they were entertained by a dedicated team of minders and play specialists in the aptly named hotel room “the Gaol”. A few breakouts were reported during the day, but all escapees were eventually returned safely to their families.

This year there was a particular focus on the positive impacts of physiotherapy and fun exercise regimes on the muscle and bone development of children and adults of all ages living with cystinosis as well as on speech and language therapy and exercises in developing speech and good throat and larynx muscles. In this context, we were delighted to welcome Christine Knerr, a speech and language therapist, and Christian Koepl, a physiotherapist both of whom work at Centre of Social Paediatrics in Traunstein Germany where they are members of the interdisciplinary cystinosis clinic which diagnoses and treats pediatric and adult cystinosis patients. Also



contributing during this session was Dr Katherina Hohenfellner who was the prime driver in establishing the interdisciplinary cystinosis consultation clinic in Traunstein which provides access to 14 healthcare professionals in a “one-stop” clinic. Christian Koepl informed us about Galileo-training programme and the use of whole body vibration training as therapeutic approach to enhance motor performance and quality of life of patients with cystinosis. Christine explained the role of logopaedic (speech and language) treatment of patients with cystinosis in improving eating and swallowing functions as well as in speech and language development.

In addition, there were highly informative talks from Professor Craig Langman (Northwestern University, Illinois, USA), Professor Paul Goodyer (McGill University, Montreal) and Dr Koenraad Veys (UZ Leuven, Belgium), all of whom gave updates on their research activities to the families and also from Elaine Mellotte, a behaviour analyst with the Central Remedial Clinic in Ireland and a trainee

educational psychologist, who focused on the difficulties experienced by children living with cystinosis and their families within the educational system.

All of the families and healthcare professionals took part in the lively, two-way question and answer session which proved to be highly informative not just for the families but also for the healthcare professionals involved.

The Annual Dublin Cystinosis Workshop is rapidly establishing itself as an important event in the cystinosis scientific calendar at which world-class international scientists from various disciplines network, share ideas, discuss scientific breakthroughs and work together with the common aim of conquering this rare disease.

This was our fifth year hosting the scientific workshop and we were delighted to welcome 35 participants including scientists and healthcare practitioners from the US, Canada, England, Scotland, Belgium, Netherlands, Germany, Italy and of course from Ireland (North and South) to Dublin. The 1½ day workshop comprised 17 excellent speakers and spanned research topics in areas such as: bone disease and

muscle wasting in cystinosis patients, genetic indicators in transplant patients, updates on new therapeutic approaches for cystinosis, a better understanding of the molecular and physiological basis of cystinosis, fertility and pregnancy for those living with cystinosis and understanding the significant social impacts of living with cystinosis. This year we welcomed a number of new scientists from research areas that are not directly linked with cystinosis but which intersect with it – Dr Olivier Govaere from Newcastle University who is researching the drug cysteamine in the context of non-alcohol related liver disease and Dr Brendan Keating from the University of Pennsylvania who is using genome-wide genotyping and exome sequencing to uncover genetic markers which may provide insight into the causes of chronic kidney disease and which may assist in the clinical management of post-transplant patients.

In addition, we were also delighted to welcome Emeritus Professor Herbie Newell to the cystinosis research family. Herbie has a distinguished research career in the development of cancer therapeutics and he has the unenviable task of taking over the research project of the late Professor Roz Anderson. Herbie provided us with a wonderful update on Roz's legacy project which focuses on the development of a cysteamine pro-drug that should provide a more effective, easier to administer treatment than cysteamine itself and with less side effects. Herbie paid a very moving tribute to Roz who passed away shortly after having attended our workshop last year. Roz dedicated her scientific life to alleviating the symptoms of cystinosis sufferers and is sorely missed by all of us who knew her in Cystinosis Ireland.

This year, as part of the scientific programme, we invited researchers, particularly early-stage researchers, to submit posters to the workshop and also to make a short oral presentation during the workshop aimed at explaining their research to the families. We awarded two prizes: the Professor Roz Anderson Prize for Best Scientific Poster and the Cystinosis Ireland Prize for Best Oral Presentation to a Lay Audience.

We were delighted to present the inaugural Professor Roz Anderson Prize for Best Scientific Poster to Amer Jamalpoor for his poster "Investigating the pathophysiology and a potential therapeutic approach for Nephropathic Cystinosis". Amer, a young and passionate researcher from Utrecht University, also won the Cystinosis Ireland Prize for Best Oral Presentation to a Lay Audience, which was decided exclusively by the votes of the families. Professor Herbie Newell presented the Professor Roz Anderson Prize to Amer. We all felt quite emotional during the presentation and very much felt Roz's presence. We want to extend a warm thank you to Roz's husband Les for allowing us to present this prize. It was truly an honour.

Hosting the 5th 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 the additional conference support received from the Health Research Board (HRB). Cystinosis Ireland would also like to thank the members of the Workshop Organising Committee – Professor Elena Levchenko

(Scientific Chairperson), Dr Atif Awan (Irish Medical Organiser), Dr Achim Treumann and Ms Anne Marie O'Dowd, with the able assistance of Dr Tom McDonald, Ms Sue Maguire, Ms Denise Dunne and Dr Ruth Davis, for all their hard work in organising this event.

### **International Cystinosis Conference 2020 – a date for the diary**

Next year, Cystinosis Ireland is looking forward to hosting the International Cystinosis Conference in Dublin on 10-12 July 2020. Preparations for the conference which will also incorporate next year's Dublin Cystinosis Workshop, are already underway and we hope to see many of you at the conference here in Dublin – so make sure you put the dates in your diary.

### **Other activities**

Cystinosis Ireland is also a part of the Cystinosis Network Europe (CNE) which comprises members from patient organisations across Europe and further afield.

A major initiative of this group is the establishment of a Worldwide Cystinosis Community Advisory Board (CAB) under the mentorship of Eurordis, the representative body for rare diseases in Europe. Cystinosis Ireland is chair of the Cystinosis CAB, and of CNE, for the next two years. The key objective of CABs is to engage with pharmaceutical companies in order to bring the patient perspective to clinical trials and other areas. Members of the CAB receive extensive education and training in all aspects of clinical trials/treatment development so that they become patient experts. The Cystinosis CAB has expanded its remit to engage at all stages of research development, including at the early research stage, and to provide advice and the patient

perspective to ensure more effective clinical trials and more efficient and speedy market access.

### **Funding Opportunities for Cystinosis Research**

Cystinosis Ireland is involved with two research funding initiatives that are open to researchers based outside of Ireland. These are:

#### **Cystinosis Seedcorn Funding**

**2019:** This fund, which is supported by Cystinosis Ireland, Cystinosis Research Network USA (CRN) and Cystinosis Foundation UK (CF UK), aims to provide researchers with the opportunity to generate solid preliminary data which would contribute to a larger, sustainable, longer-term application for funding.

Cystinosis Ireland is particularly open to new, high-risk, blue-sky applications and will consider these favourably (Applicants should highlight this in

their application).

Typical projects funded tend to be in the region of €10,000, however, the Cystinosis Ireland executive will consider applications that are for more than this (even substantially more) where the project is well justified and is relevant to patients and families living with cystinosis. All research proposals are subject to peer review.

Closing date for applications in 2019: 2 August 2019; 29 November 2019

Researchers interested in this scheme should email [research@cystinosis.ie](mailto:research@cystinosis.ie) for an application form and to discuss any potential applications.

#### **MRCG-HRB co-funding scheme**

**2019.** Cystinosis Ireland is very interested in supporting high quality research proposals to the MRCG

HRB co-funding scheme.

The next call for proposals is expected to open in September 2019.

Cystinosis Ireland has a strong track record of supporting research proposals for funding through this co-funding scheme and currently has two active projects underway.

MRCG HRB awards are typically in the region of €100,000 per annum for up to 3 years.

Closing date for applications: The deadline for submissions is not yet public but is expected to be prior to Christmas 2019.

We encourage researchers to contact us at [research@cystinosis.ie](mailto:research@cystinosis.ie) at any time in order to discuss your research idea to see if it might be suitable for this funding scheme.

## **Jack Greeley 19th Birthday Letter Fundraiser for CRN**

*By Christy Greeley, Vice President of Research*



*Stevenson HS Class of 2018 with Alex.*

The Greeley Family once again celebrated Jack's St. Patrick's Day birthday sending out the 17th 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. 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 nearly \$400,000 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:

How in the heck did Jack Greeley

become a college freshman and 19-year-old? Those that know Jack's story can marvel at these accomplishments with us.

For 17 years now, we have been updating our "circle" about our son Jack who on many levels is a marvel in his own right whether past, present, or future. As most of you know, Jack has cystinosis, a rare metabolic, genetic disease affecting about only 2,000 people worldwide. Christy runs the Cystinosis Research Network (CRN), an internationally recognized 501(c)3 aimed to assist the fight with this affliction. As featured in CRN's most recent newsletter (Fall/Winter 2018), there was a research grant CRN funded for a study out of Belgium that offered a good broad

strokes framing of cystinosis: "...a genetic disease manifesting early in life (~ 6-12 months) with progressive kidney disease resulting in renal failure 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."

Like life itself, cystinosis and Jack offer plenty of "unknowns," yet many "knowns" as well. This year's letter is aimed at giving a simple and general update on Jack; both those known and unknown variables of our Champ. What have we learned or what do we know about Jack over the past year? The list is plentiful and wonderful:

- Jack is over halfway through his freshman year at Marquette University, a Jesuit school in Milwaukee that seems to be a great fit for him – close to the home front, a compact campus, and a strong support and curriculum system.
- He entered college like the typical first-year student needing to make a transition and adjustment. Scholastically, he started with course work that included Biology and Chemistry, which we quickly determined was too heavy a load. Hmmmm, if not Biology, what should Jack major in now? Anthropology it is!
- He joined the E-sports club and became a part-time student manager for the Golden Eagles varsity program, which put him in contact with two loves, video games and basketball, respectively.
- Jack has the usual adjustment of any college freshman taking care of himself, but his was a bit more



*Marquette Parents Weekend.*

involved, including taking over his strict regimen managing his own medications, which includes taking over 30 pills a day split into four separate time periods. This was a major milestone and breakthrough as adhering to his medicine will make an immeasurable difference for him and his future. At Christmas break, we realized that this would need to be tightened up because he came home with too many pills. The routine is vigilant, but the math is simple.

- Like all college kids, Jack was now on his own with personal hygiene and wellness. Showers are not really the issue as much as muscle weakness, which results from cystinosis. With years of occupational therapy in his background to help with fine motor skills, he was forced to clip his nails by himself (yes, a simple task, but a milestone). He has done a really nice job there, but if only he would remember to go to the barber every four weeks and occasionally shave. Ha!
- Being on campus, Jack was forced to walk more than his surgically repaired legs might be accustomed. The good news is that as his legs got stronger, his walking became more upright. He – not Mom, not Dad – now more liberally decides when to wear his leg braces or not.



*Guinness Storehouse, Dublin.*

- As a pretty big guy pushing 6-1 and 200 pounds, Jack lost some weight, which we theorize came from a busy schedule and – being a tad forgetful – came from simply forgetting to eat. Oops, the unlimited, 24/7 meal plan was optional!
- Not a heavy socializer or partier, we learned something great about Jack that came to light because of an equally great law in Wisconsin where an 18-year-old can drink legally with his parents. Being close to Jack, we have enjoyed some dinners with him in Milwaukee (about an hour from the house) and he has taken the liberty of ordering a hard cider, which sounds like a chip off the old parental block. Like any college freshman, this list is pretty common and completely normal, just like Jack, short of his medical challenges. But what about the unknowns?
- How will we track his daily medication routines? Eight-plus iPhone alarm reminders is certainly intended to help.
- Due to his kidney disease, Jack is a prodigious water drinker. He swears he has never locked himself out of his dorm room when he gets up every night at 3 am to take his meds and go to the bathroom across the hall. The good news is that we have never

gotten a call in the middle of the night – for a lockout or an emergency.

- How can we be of service to Jack? The thing about Jack is that he rarely asks for anything and he never, ever complains. Are we doing everything we can?
- Will he admit if there is an issue? Jack probably suffers from foolish pride where he wants to be an adult and is hesitant to ask for assistance.
- To this day, we marvel at how “chill” Jack is and we wonder how that happened. Will he even be able to recognize when he might need help and would he even tell or ask us? How do you tow the parental line of being loving and caring without being a total pain? Jack has been picked, prodded, poked, cut, examined, nagged, etc. so much over his life that we try to find that balance as he matriculates into adulthood and not just college.
- During each school break, we

schedule appointments with a bevy of medical specialists that help take care of Jack – pediatrics, orthopedics, nephrology, gastroenterology, ophthalmology, genetics, cardiology, neurology, pulmonology, and endocrinology. How do we know what those tests and exams will show... and what might be a result of him being on his own as he establishes his independence?

- Last Spring 2018, Jack and our family participated in a study at the NIH’s Undiagnosed Disease Program for an examination of possible new causes of cystinosis, given Jack’s unique orthopedic challenges. He could not have better care, but what might this new research show?
- What if something goes terribly wrong at school? How will Jack be cared for immediately?
- Like all parents, we think we have planned and prepped Jack appropriately, yet doesn’t hope remain

one of our biggest allies?

We all know about life’s trials and tribulations. This applies for anyone – whether completely healthy or not. When dealing with a rare disease like Jack has, we stopped asking long ago, “Why Jack?” It serves no purpose and we ceased doing it. There is a great scene in the Academy Award winning movie, Unforgiven starring Clint Eastwood as grizzled, reformed gunslinger William Munny. He is forced out of retirement to take on iron fisted and exploitative Sheriff “Little Bill” played by Gene Hackman. In the film’s climax – spoiler alert – Munny is hovering over the dying Little Bill, who claims he doesn’t deserve to go out this way, when Clint corrects him before finishing the job by grousing, “Deserves got nothin’ to do with it.” While Jack does not deserve his plight with cystinosis, he does deserve opportunity and the greatest future we can help provide.

## Resources at Your Fingertips: New Cystinosis.org Website

This April, the cystinosis.org website revealed a new look. Inspired by community feedback, the site has been updated to include pertinent information just a few clicks away. Please take a moment to browse around. Here are some ideas on where to start.



**Where are you in the cystinosis journey?** From initial diagnosis, managing

daily life and/or navigating adulthood, there is something for everyone within the About Cystinosis and Support & Resources sections.



**Translation services.**

Powered by Google, our website information can be translated into over 40 languages.



**Accessibility options.** By selecting the stick figure icon on any page, you can alter the contrast, text size, highlight links and have content read aloud.



**Publications and Guides.** Review a list of articles by publication date or search by category or author. (See Support & Resources section)



**Visit News & Events** to catch up on what’s happening within the cystinosis community.

# Wyman Facebook Fundraiser

By Jen Wyman, Vice President of Family Support

It always astounds us...the family, friends and strangers who read our story and give so generously. It is never, ever easy to ask people to give away their money. But when you help them to see how the cause and the organization they are supporting gives back to it's community they do it willingly and happily. We are grateful for everyone who donates each and every year without hesitation. In the giving of their dollars they also give a tremendous amount of hope.

Following is the Facebook fundraiser letter celebrating Kacy's 4th Anniversary with her dad's kidney. It has generated over \$9,000 to date. We hope to hit the \$10,000. Our family will donate a matching \$10,000.

It's that time of year again...when I unabashedly brag on Kacy while simultaneously ask for your generosity in donating to The Cystinosis Research Network.

It's been 4 years, 40,000 pills and 4,000 eyedrops (give or take) since her kidney transplant. She takes her medications 4 times a day and visits clinic 4 times a year. (I do believe we need to play 4 in the lottery). On a serious note however, this will probably never change. While her kidney transplant has improved her life, she is not cured of cystinosis.

With every year she will face new and different struggles. She will never live without medications.

Her compliance and diligence in living with cystinosis and caring for her kidney is the number one reason why she remains stable and healthy. That said, it is no easy task.

It could be that living with a rare disease and following a strict, time sensitive medication routine has led to her "super"organized lifestyle. Or it's possible that her organization and independence are why she is able to care for herself so well . Either way, her health, her grades and her work ethic are proof of that one compliments the other...the order and the reasons don't really matter. She will be 17 this summer and is without a doubt growing into her adulthood. We couldn't be more proud of her ability to "juggle" her life.

She is completing her sophomore year at Bloomfield High School and works 25+ hours at Beyond Juice (during the school year). She will attend the Solstice Teen camp NorthStar Reach again this summer, work full time and take a summer civics class. She is happy and healthy. It's all we can really ask for.

We will be in Philadelphia for the Cystinosis Research Network

conference in July. It is a highlight of her summer, reconnecting with friends who share the same struggles and learning how to better care for herself as an adult living with the disease. We are fortunate to have this organization to support and guide us on the cystinosis journey.

SOOOOO... If you find yourself searching for a place to spend your charitable dollars or you just feel like throwing a few our way I hope you will consider CRN. I promise you that your donation is 100% used in the best ways possible...on cystinosis education and research and continually and directly making Kacy's quality of life the best it can possibly be.

You can learn more by visiting our newly designed website at [www.cystinosis.org](http://www.cystinosis.org).

Thank you for supporting us in every way possible. It has been 13 years since diagnosis, but a lifetime for Kacy.

If online donation is not your thing we would happily accept a check made out to the Cystinosis Research Network (in honor of Kacy) send to the Cystinosis Research Network, 302 Whytegate Court, Lake Forest, IL 60045 USA.

*"There is no exercise better for the heart than reaching down and lifting people up." — John Holmes*

# Cystinosis Day 2019: Making a Difference, One Step At A Time

May 7th marked the 2nd Annual Cystinosis Awareness Day.

The date, 5/7, was chosen to represent the most common cystinosis mutation: 57-kb deletion. In two short years, this day has been met with an abundance of support from around the globe. This year it was honored by over 20 countries, reached over 30,000 people online with fundraising efforts exceeding \$25,000. Above all, you've help transform an ordinary day into one filled with education, advocacy and contributions benefiting our rare disease. Thank you!

The pride and inspiration Cystinosis Awareness Day has created does not need to stop on May 7th. Please continue to commemorate this occasion. Ideas on how to get started can be found at [cystinosis.org](http://cystinosis.org) under the How to Help tab. You can also donate directly at [cystinosis.org](http://cystinosis.org) or mailing checks written out to the Cystinosis Research Network to 302 Whytegate Court, Lake Forest, IL 60045 USA.

To catch up on events from the inaugural Cystinosis Awareness Day, check out our Inspirational Stories page: <https://www.cystinosis.org/>

[our-impact/inspirational-stories/](#). With his 2018 efforts considered a "successful failure," Clinton Moore (CRN President), challenged himself and a small group to walk non-stop for 57 miles in less than 24-hours. In addition, the Adult Leadership Advisory Board (ALAB) recruited community members to walk, run or hike a collective 57 miles. Continue reading to see how their events turned out.

**Planning for Cystinosis Awareness Day 2020 starts soon!** For information, email Clinton at [Clintonmoore1@aol.com](mailto:Clintonmoore1@aol.com).

**How rare is cystinosis?**

An estimated 2,000 people are living with cystinosis; occurring 1 in every 100,000 - 200,000 live births.

#cystinosisaware

**Is cystinosis just a kidney disease?**

**No.**

Cystinosis impacts every single cell in the body.

#cystinosisaware

## Rare Disease Day 2019: Awareness for #MyCystinosis

On the last day in February, communities come together to raise awareness for the over 7,000 known rare diseases. This year, the CRN launched a temporary tattoo awareness campaign. Free of charge, tattoos were shipped to all registrants completing the online form. Once received, they were encouraged to share an applied tattoo photo with

a piece of their cystinosis story via social media (#MyCystinosis) and/or within their daily lives. Requests were received from around the globe with hundreds of tattoos paired with advocates in 22 countries!

Thank you to those near and far who made the campaign a success by "sharing their rare" before, during and after Rare Disease Day!



# 57 Mile Walk for Cystinosis Research Network

By Clinton Moore, President



Planning for this year's 57 mile walk started roughly 2 months before the event. The option to hold it at the same location as last year was a possibility but I wanted to change things up a bit. With 12 people wanting to join me for every step, I wanted to somehow make the walk resemble the life of a cystinosis patient and caregiver as much as possible. To show them a little if what it's like to walk in our shoes. How to do this and tie it to a walk I found was quite easy.

The venue chosen was a local high school. On the track behind the building that is only a quarter mile long. This is going to mean that 228 laps around this small track is needed to reach 57 miles. Ridiculous right? Exactly!!!

Training began and for me this meant working all day and then trying to get in a lengthy walk after work and before dark every day. Didn't always happen but was able to get it in at least 4 or 5 days a week. Some days I only had time for a few miles and others I could go quite a bit farther. This was my routine for a couple weeks and then a slight complication surfaced. Without getting into great

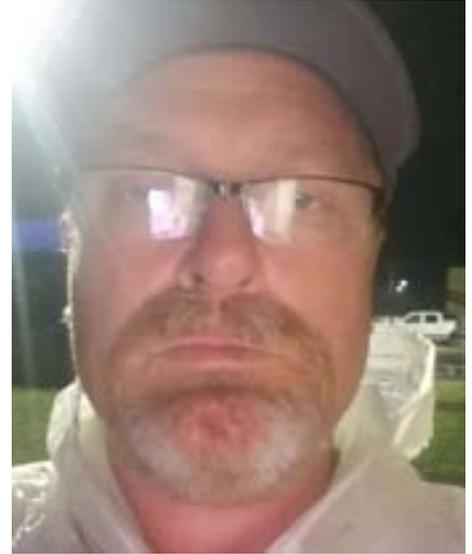
detail, my son had an unknown health issue that suddenly required me to be home quite a bit more than before, so my walk schedule became more and more scarce. Did what I could as much as I could and when it became walk day I had totaled right around 250 miles in training. Hope it was enough.

Walk day came and although you've walked what seemed like an eternity already, you can't help but wonder if you trained enough. My team all arrived and after a short pep talk we were ready to begin at noon sharp.

First few miles and hours are always easy. You're fresh, rested, and plenty of people around to talk to. I know that will change but I'm not sure the others on my team know what's coming. What they don't know is that every detail of this walk in some way resembles my life, my son's life, and every life out there that is affected with cystinosis.

Before dark, the walk begins to take its first casualties. Blisters and hurting feet begin to force individuals to sit down and be unable to continue. I hear people saying that walking the same lap over and over is getting





monotonous. I overhear others saying this is too repetitive and should of been held at a place with a longer loop. I see small groups forming as they walk and either get way ahead or way behind the others. 1 by 1 I watch my team fall into the wrinkles of this walk that I intentionally put in place.

Night comes and amongst the darkness, I slowly see my team deplete. Some with a hand shake and a smile, others with tears streaming down. Unfortunately for the ones remaining there is another part of this walk that is about to become a reality that even I couldn't control but love how it ties in with all the other

obstacles. Rain. Light at first then becoming quite steady. Now when I look around the track, I can't tell who is who as they all look the same with ponchos on. Everyone that remains is doing what I knew they would be doing. I call it stare walking. Few people are talking, and everyone is staring at their feet putting one foot in front of the other. Now is the time I believe no one is having fun. It's miserable out here. Puddles are forming so you have to be more conscience of where you walk. The rain is chilly. Sunrise is still hours away and there is still many laps left to go.

So let me back up a minute. How does all this resemble the life of someone affected with cystinosis? How can a walk resemble that at all? Well...that's easy.

1. Upon diagnosis, you enter into an unknown world. A life you're not familiar with. A life you know is going to be full of challenges but you don't know what they are or when they will occur. This walk gives the same beginning. Entering into this walk for your first time you don't know what's to come or how you will handle it. You don't know the challenges both physical and mental that are to come. You just know it's going to be a challenge.

2. Cystinosis is a very repetitive disease. Staying compliant and as healthy as you can be requires an internal time machine as you need to take meds at the same time every day. Doctor visits on a regular 2 or 3 month schedule. Everything repeats itself. Over and over and over. Days after day. Week after week. This walk is no different. 228 laps around a small track is very repetitive. Keep walking the same lap over and over and over. No matter how repetitive it is...keep going.



3. Cystinosis can be very monotonous. Doing the same thing over and over and over can drive some people nuts. To the point they need a break and want to stop doing the things that keep them healthy. Again...the walk shows that. If 228 laps isn't monotonous then you're a very relaxed person.

4. Storms. Cystinosis can suddenly bring on challenges without warning. Health issues can occur with little more than a minutes notice and you push through. Take the necessary steps and keep going. So remember when it began to rain in the middle of the night? Unplanned, not scheduled, unwanted, yet unstoppable.

5. You know those times when you suddenly have to head to the hospital for whatever reason, you go through the ER and sit there for what seems like half your life, just to then realize you're getting admitted...and then you wait the other half of your life for an available room? Then when you finally get to your room and get settled you realize how hungry you are so you call the cafeteria just to realize they are closed for the evening?? So you end up settling for just the snack machine to keep you alive until morning?? As terrible as this sounds... we also did this to our team. We gave

them food in the early hours of the walk...but after that when they were exhausted they had a table of just non satisfying snacks to get them through till morning.

6. Cystinosis can become exhausting. When you keep up daily with everything involved it certainly can be exhausting. Again...228 laps certainly does too.

So...now that you know a few of the details on how this was all planned out, it makes a little more sense now huh?

Sunrise finally shows up and there is myself...along with 2 others from my team that has been on that monotonous, exhausting, repetitive, rain covered track, nonstop, without leaving or resting, since we started. The end of this 57 mile journey on a quarter mile track is nearly over.

As in the beginning, now that we near the end, many people have shown up to cheer us on and walk that final lap with us. Many people are getting emotional, myself included, as we round that last and final turn. It's the turn we've wanted for so many hours. And it's finally behind us.

Thank you to all of you that participated, supported, and donated to this wonderful organization during

this event. We look forward to next year and yet again...grow participants and make it more challenging than ever.



# 57 Mile Walk for Cystinosis Walk - ALAB Edition

By Heidi Hughes



*Shannon Keizer and team.*



*The Healy twins, Sara and Jana.*



*Heidi Hughes.*



*Eddie Langley, Jr. with family and friends.*



*Cheryl Simoens, our Canadian representative, and her companion.*

*30 participants. 160 miles completed.*

330,000 steps. This is the distance members of Cystinosis Research Network's new Adult Leadership Advisory Board (ALAB), our fellow cystinosis warriors, and their loved ones walked for 2019 Cystinosis Awareness Day.

Helen Keller said, "Walking with a friend in the dark is better than walking alone in the light." CRN's President, Clinton Moore and his mighty team proved this by persevering for 22 hours and 57 miles through daylight and darkness with the entire cystinosis community as their cheerleaders. ALAB joined forces as our inaugural event to raise awareness, spread hope and the message of strength for all our fellow warriors while tag teaming the fundraising efforts. Collectively, this community exceeded the fundraising goal of \$5,700 and demonstrated that together we can do anything, and we will all rise above this disease one step at a time.

Throughout the past 6 months I have hiked hundreds of miles throughout the state of North Carolina during the recovery from my second kidney transplant. During one of these adventures, the thought of Clinton's '57 Miles for Cystinosis Walk' and his dedication towards Chandler's future and our entire community motivated me to brainstorm ideas to use my treks for good. But how? Could my recovery walks team up with Clinton's famous 57 miles and make a difference? The answer was in the

sweat dripping down my forehead and excitement growing with every step. Yes, however, I needed to recruit more soldiers. I decided to propose the idea to my ALAB teammates to collaborate with Clinton's walk. Everyone enthusiastically agreed and jumped on board right away committing their ambitious distances to our 57-mile goal. I knew with all our efforts combined we too could travel the same distance across North America as Clinton and multiply our impact. So, we did! The '57 Miles for Cystinosis Walk – ALAB Edition' Facebook event was created, and it spread like wildfire as our mileage and involvement increased quickly every hour.

Together my ALAB team, fellow cystinosis warriors, and our loved ones dedicated over 160 miles with over 30 participants on trails, treadmills, and rock walls and exceeded my expectations for this event. Each drop of sweat and every mile walked not only made us stronger as a community it also got us closer to a cure and many miles further in our journeys through life with a rare disease. I am so grateful for everyone who got out there, got active, and spread awareness.

Thank you all for your participation and remember to stay strong and never stop marching towards the brighter future for cystinosis.

P.S. Start training now for our 2020 57 Miles for Cystinosis Walk – ALAB Edition!



PROCYSBI®  
Empowering Patients  
With Cystinosis



Horizon Therapeutics 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 2019 are Pittsburgh, Pennsylvania; Kansas City, Missouri; Cleveland, Ohio; Minneapolis, Minnesota; Boston, Massachusetts; Charlotte, North Carolina; San Antonio, Texas.

**To RSVP to one of these cities or to request a program near you,  
please call 602-953-2552.**



Horizon Therapeutics 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** 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](http://PROCYSBI.com).**

# FAMILY STORIES: The Gustafson Family

By Amanda Gustafson



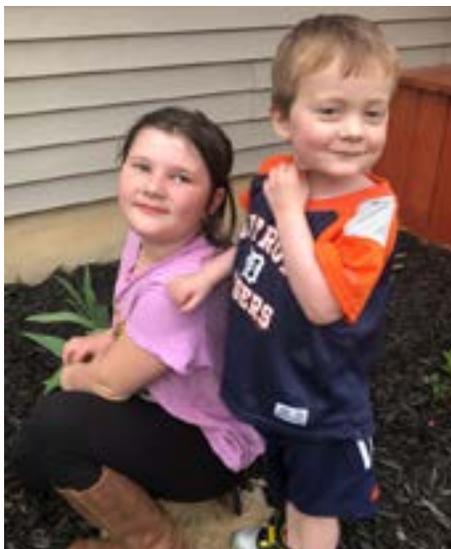
*Super Gavin and the Great Pumpkin Patch.*

Gavin was diagnosed with cystinosis at 6 months old. When he was around 5 months old he stopped drinking his formula as much, and started to get constipated and spit up more often. We took him to his pediatrician and they sent us home stating he had reflux. When the reflux meds weren't helping and the constipation was too bad we finally went with our gut and took him to the ER. They didn't do much besides an abdominal x-ray and sent us home. We went back to the pediatrician the next day and she ran STAT labs and a urine sample. I remember clearly that we had a birthday party that day and as we were on our way, planning to have a day of fun I got a call to go to the ER immediately instead because he had glucose in his urine and other lab issues. Our heart and stomach dropped. In the ER he had many tests run and every specialist we could think of came in to see him, trying to figure things out. Finally an amazing nephrologist, Dr. Saberi came in. He told us he thought Gavin had a rare genetic disorder called cystinosis but it would take awhile to confirm because the blood test had to be sent out. He also told us he had Fanconi



*Our little man.*

Syndrome. I never googled so hard in my life, as I had never heard of it and after reading article after article I was like, this is what he has, this is it. Once we got his levels on track we were sent home with medications and told to wait. We followed up with the nephrologist in July 2014 and he confirmed the diagnosis of cystinosis. We were devastated, but eventually adjusted to our new normal. He had to get an NG tube shortly following diagnosis due to weight loss and then finally a G-tube to help with his medication and food intake as he would not eat/drink much orally. We



still struggle with weight and growth but we are getting there. Gavin is now 5, and the happiest, most loving kid in the world. His big blue eyes and smile light up the room. He has had a few hospitalizations since diagnosis, but not many. He is a goof ball who beats to his own drum, our little wild bear cub. He loves telling stories, Minecraft, LEGOs, Superheroes and just being a crazy kid, He loves his big sister Sophie, and his two dogs and cats, and of course mom and dad. He is so full of joy and happiness that he makes everyone he meets smile. He is starting Kindergarten in the fall and is very excited! We have good days and bad days, just like everyone else, but we try not to dwell on the bad. We live every day with gratitude and don't take a single moment for granted. He has cystinosis, but he is not cystinosis, we won't let that be his identifier.



## FAMILY STORIES: The Sheperd Family

# In rare San Antonio transplant, brother and sister each received a kidney from same donor

May 6, 2019 | San Antonio Express-News | By Lauren Caruba | <http://bit.ly/2MZR5oL>



*Dr. Elizabeth Thomas, left, associate professor of surgery at University Transplant Center, talks about the unusual double kidney transplant that gave one organ to Ava Shepperd, 14, hugging her father John Shepperd, and the other to her brother John Ben Shepperd, far right, next to his mother Kim Azar Shepperd. The brother and sister both have a genetic disorder that compromised their kidneys. The kidneys came from one deceased donor. The transplants took place at University Transplant Center on May 3, 2019. Photo: Bob Owen /Staff photographer*

For years, John Shepperd and Kim Azar Shepperd have known both their children would eventually need kidney transplants.

The siblings, Ava and John Ben Shepperd, were born with cystinosis, a rare genetic disorder that causes a type of amino acid to accumulate in the cells. The once-fatal disease, which affects several hundred people across the country, can damage multiple organs, particularly the eyes and kidneys, necessitating transplants early in life.

The family had joked that all they needed was one donor who could provide kidneys for both children. They never anticipated that it could actually happen.

Last week, their mother received a call from University Transplant Center,

where 14-year-old Ava and 18-year-old John Ben were listed as kidney transplant candidates. When she was told there was a potential donor for her daughter, she asked about her son, who had nearly died as a baby before he was diagnosed.

Both were a strong match.

After frantic calls between the two halves of the family — Ava lives with her mother in Alpine, John Ben with his father in Austin — they packed their bags and made separate drives to San Antonio.

On Friday, within hours of each other, the teenagers underwent transplants at University, each receiving a kidney from the same deceased donor. The surgeries overlapped by about an hour — “the longest and shortest hour ever,” their father said.

Days later, the family is still reeling from the extraordinary turn of events.

“Obviously, we never thought it would happen on the same day, from the same wonderful donor,” said John Shepperd, 50. “We truly feel blessed.”

Several physicians at University Hospital were also astounded.

“I’ve never seen this before, in my 10 years of medicine,” said Dr. Daniel Gebhard, who works in pediatric intensive care at the hospital.

A successful kidney transplant requires not only the same blood type, but also compatible antigen typing and cross-matching.

Moreover, a computer program is responsible for matching potential donors with organ transplant candidates. Depending on the circumstances, an organ may not necessarily go to the nearest transplant center and could be transported elsewhere. Adults in South Texas usually spend years waiting for a kidney transplant. Pediatric patients are given higher priority.

In light of those factors, last week’s outcome was “incredibly rare,” said Dr. Elizabeth Thomas, a transplant surgeon at University Transplant Center. “The computer did it for us.”

Also, the donor’s liver was split and transplanted to two recipients, which is less common, Thomas said. The donor has not been identified due to privacy laws.

For Ava and John Ben, who both needed dialysis, their surgeries came as a relief. John Ben, who has

suffered from more complications from the disease, has undergone many surgeries.

This one felt different.

"I remember hooking up to my dialysis unit and thinking, this is the last time I'm going to have to attach this dumb tube to my stomach," said John Ben, who will graduate this month from Austin High School. "It just felt really good to know that this is the last time I was going to be doing this."

Along with warding off organ rejection, Ava and John Ben will still have to manage their cystinosis. But the disease will not attack their new kidneys, their mother said.

On Monday afternoon, the Shepperd family wore surgical masks to protect the teenagers' immune systems, made vulnerable by immune-suppressing drugs, as they answered questions from reporters in a hospital room.

Ava, who attends seventh grade at Alpine Middle School, said the attention made her nervous, and the procedure itself was daunting. But she said she has been cheered by the support from her friends and school.

Thomas said the situation shows that "there is no normal transplant."

"Every donor is very unique in the way they touch lives," she said.

Lauren Caruba covers health care and medicine in the San Antonio and Bexar County area.

Twitter: [@LaurenCaruba](https://twitter.com/LaurenCaruba)



*Ava Shepperd, 14, left, is interviewed as her mother, Kim Azar Shepperd, right, listens in her room at University Transplant Center on May 6, 2019. Ava and her brother, John Ben Shepperd, 18, who both have a genetic disorder that compromised their kidneys, were recipients of kidneys from one donor. Photo: Bob Owen /Staff photographer*



*Ava Shepperd, 14, rests her head on the shoulder of her father, John Shepperd, during interviews May 6, 2019, at the University Transplant Center, where she and her brother John Ben Shepperd, who both have a genetic disorder that compromised their kidneys, were recipients of kidneys from one donor. The transplants took place May 3. Photo: Bob Owen /Staff photographer*



*Ava Shepperd, 14, left, and her brother, John Ben Shepperd, 18, second from right, visit with their parents Kim Azar Shepperd and John Shepperd, right, on May 6, 2019. The brother and sister share a genetic disorder that compromised their kidneys. They had been on dialysis until each received a kidney from the same donor on May 3, 2019. Photo: Bob Owen /Staff photographer*

## FAMILY STORIES: The Sheperd Family

### Siblings Receive Kidneys From Same Donor on Same Day

May 6, 2019 | Rivard Report | By Roseanna Garza | <http://bit.ly/2Flwdxh>

Two siblings with a rare genetic disorder received life-saving kidneys from the same donor on Friday, an experience as rare as the diagnosis, according to specialists at University Hospital, where the transplants took place.

Ava Shepperd, 14, and her brother John Ben Shepperd, 18, suffer from cystinosis, a lifelong condition diagnosed in childhood – typically by 6 months of age – that slowly destroys the organs in the body, including the kidneys, liver, eyes, muscles, and the brain, due to abnormal accumulation of the amino acid cysteine.

Only 500 people in the United States have this diagnosis, and 2,000 people worldwide, said Dr. Elizabeth Thomas, pediatric critical care specialist with University Hospital. An estimated 20 new cases of cystinosis are diagnosed each year.

“It is exceptionally rare [to have siblings with the same diagnosis receive needed organ donations] on the same day from the same person. Kidney allocation is pretty complicated. It’s a ranking mostly dependent on how long someone has been on dialysis. So the fact that these kids both had their name come up at the same time and they were both a match for this donor is very unique,” Thomas said.

John Ben was diagnosed with cystinosis at 14 months, while Ava was diagnosed in utero, said their mother, Kim Azar Shepperd. Since then, both had to take handfuls of medications per day to regulate symptoms and stave off kidney failure.



*(From left) Ava Shepperd, Kim Azar Shepperd, John Ben Shepperd, and John Shepperd talk about Ava’s and John Ben’s transplant experience after both received kidneys from the same donor.*

“We have known since the beginning that they would both need kidney transplants, but there was no way of knowing when that would be. And these kidneys are basically perfect, more than we could have ever hoped for.”

Ava and John Ben had been on dialysis for kidney failure for extended periods of time before the organs became available — Ava for six months and John Ben for more than two years.

John Ben told the Rivard Report that he is excited to receive a kidney ahead of graduating from high school at the end of the month.

“I did all of my dialysis at home at night, and I had to be home for the entirety of the night because it took eight hours to complete,” said John Ben whose dialysis was required daily. “This started my sophomore year in high school, so it was a little troublesome.”

Seventh-grader Ava completed

dialysis at least three times a week, with each session lasting over three hours. “Being on dialysis taught me to value my friendships, because I missed out on so much at school and with my friends because I had [to complete treatment],” Ava said.

The family got word at 7 p.m. Friday that both siblings would receive kidney donations from a deceased donor who was a 92 percent match, after spending more than a year on the transplant list for John Ben and three months for Ava.

Because an organ transplant can only be given to a healthy recipient, John Ben had been unable to receive an organ on three separate occasions when his name came up because he was very sick at the time, which is a big reason they received them at the same time, Azar Shepperd said.

Ava lives with her mother in Alpine, while John Ben resides with his father, John Shepperd, in Austin. Both were registered on the University Hospital

transplant list, which does not require a recipient to live in San Antonio as long as they meet organ donation criteria, Thomas said.

“If you meet the criteria we will register you to receive an organ. If no one on our list needed these kidneys,” they would have been made available first regionally, then nationally, to those who did, Thomas said, noting that children are prioritized on organ transplant lists “because that’s how it should be.”

To take care of their new kidneys, the siblings will take multiple daily medications to ensure that the new organ remains healthy as it acclimates

to the body, said nephrologist Ikuyo Yamaguchi. “But the exciting part about this transplant is that it took place just three days ago and these kids are doing very well immediately after.”

The entire transplant experience took less than 24 hours between calls made about the organ availability to the organs being transplanted in each child, Shepperd said.

“We never in million years would have guessed that they would be receiving kidneys on the same day at the same time. It was an amazing experience and an amazing testament to the importance of kidney donation

programs and organ donation programs around the country. We feel truly blessed and extremely fortunate,” Shepperd said.

Ava and John Ben will have to follow up with doctors for one year to make sure that the organs remain healthy and are not rejected by their bodies. Because of this, John Ben plans to attend Austin Community College before moving to Colorado to pursue a degree in information technology. Ava will start eighth grade at Alpine Middle School, where she looks forward to having a “more normal experience.”

“I already feel better and healthier, like I can do more,” Ava said.

## The Patient Congress – March 11 & 12, 2019

**HILTON PHILADELPHIA AT PENN’S LANDING** — Clinton Moore and Marybeth Kruppenacker attended the World Congress meeting, recently held in Philadelphia. This meeting was an opportunity to listen to and participate in various breakout sessions and hear from some of the leaders in patient adherence and engagement, patient advocates and about rare disease market access. Meetings like these allow Cystinosis Research Network the opportunity to build on partnerships with industry leaders, government regulators as well as network with other patient groups. The breakout sessions are run in such a way that attendees are encouraged to participate and

ask questions as well as have informal discussions with other Rare Disease Advocates. As part of the Patient Advocacy Meetings, there were discussions on how to develop and build mutually beneficial partnerships to meet the needs of patients. Additionally there were speakers who spoke about developing outcomes and processes to measure the value and impact of advocacy initiatives. The value of building on those partnerships and how to improve access and affordability, in addition to how to involve patients in public policy initiatives and the critical importance of the value of that involvement. Health care stakeholders are in the midst of radical change

and organizations must focus on managing costs, improving and enhancing patient experiences. A big topic of discussion was the importance to identifying the needs of the patients and incentives to continue to work together and collaborate and improve on patient outcomes both within industry and government. The changing health care markets are becoming more and more difficult to navigate and information shared at meetings such as this helps organizations like CRN to learn what are the best and most practical practices and how to become the best advocates for all.



 <b>200+</b> <b>ATTENDEES</b> Expected	 <b>75+</b> <b>SPEAKERS</b> Including Chief Executives & Thought Leaders	 <b>35</b> <b>SESSIONS</b> Keynotes, Panel Discussions, Case Studies, & Interactive Breakouts	 <b>2</b> <b>Days</b> Engaging content & networking
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## FAMILY STORIES: Cystinosis: A Blessing and Burden

By Steve Schleuder

In order to think of what Cystinosis Awareness Day means to me I had to think about what cystinosis means to me. Cystinosis is a blessing and a burden. The blessings of it are not obvious growing up. There is physical, physiological, emotional, and spiritual scar tissue that forms and has to be acknowledged and confronted in order to be able to fully move forward with life. However, when looking back at my life with cystinosis, I've realized that without having it I would never have met some of the people I have in my life. I am more empathetic towards a wider variety of people than I think I would be if I hadn't experienced what I've experienced. I've been able to develop patience and adaptability when it comes to difficult and joyous life circumstances. I've had to mature faster in some ways than I would have had to without cystinosis, but I'm thankful for my experiences because they've made me the man I am today—and I like I who I am. However, the persistent emotion I still struggle with

on occasion is loneliness.

I've struggled a lot with the psychological and emotional issues of having a rare disease. I've been lonely and put up unnecessary walls between myself and other people. When I was a teenager, I rejected some relationships as a preemptive strike. I rejected them before they could reject me. To this day I occasionally still catch myself doing this. I have to consciously remind myself to give people a chance. I think this is why I've been reluctant to be completely open about having cystinosis with my non-familial relationships. Part of me still thinks that if I tell someone about cystinosis they will either define me solely as the guy with a rare disease or reject me outright. I know this is not the case on a conscious level, but I still have to train myself to fully accept the real truth.

I'm getting to the point now where if someone asks why I take the pills I

take, I can be completely honest with them about cystinosis. When I've revealed this information about myself the reaction most of the time is shock at what I've been through and interest in what cystinosis is. Awareness of cystinosis is key to helping more people be treated and eventually cured of cystinosis. As a healthy adult with cystinosis I am well qualified to help spread awareness. I need to fully embrace my identity as an adult living with cystinosis to help spread awareness.

Cystinosis Awareness Day means having more doctors and people know about cystinosis. This will lead to more patients being diagnosed earlier and hopefully lead to a better prognosis. Increased awareness will also help us find a cure quicker. Most of all for me, Cystinosis Awareness Day creates the opportunity for us to find people and families with cystinosis and tell them that they are not alone.



*Steve from infancy (left) through adulthood (right).*

# CRN Exhibits at Pediatric Academic Societies 2019 Meeting

The historic Baltimore Harbor was the backdrop for this year's Pediatric Academic Societies (PAS) 2019 Meeting held April 27th – 30th. CRN once again participated along with over 150 other professional organizations exhibiting in the medical field including clinical researchers, hospitals and university medical centers, pharmaceutical industries and many more. This particular event is especially rewarding as we have exposure to all the specialties in the pediatric field of medicine in addition to nephrology. It gives us the opportunity to expand their knowledge on the most up to date research and information on cystinosis. Our

new display was unveiled by Christy Greeley, Marybeth Kruppenacker and Carol Hughes. In addition there were USBs distributed to attendees that were preloaded with 5 publications on cystinosis; Cystinosis Parent Handbook, Standards of Care, Transition Management Tool, Transition Guide for Teens and Young Adults, and Cystinosis: The Evolution of a Treatable Disease. These articles are also available on our newly updated website ([www.cystinosis.org](http://www.cystinosis.org)) to review and download.

Many physicians that care for cystinosis patients as well as several of CRN's Medical and Scientific Advisory Board Members personally

visited our booth during the three days of meetings. It is always rewarding to represent the cystinosis community and have the opportunity to speak to physicians from around the world about cystinosis and share our experiences with them. Doctors are always complimentary and impressed by our presence at this meeting. CRN has exhibited at this annual meeting for more than 15 years and look forward to continuing our relationship with PAS and attending next year's summit in Philadelphia.

Christy and Carol even found time to support the PAS 5k Run for Pediatric Research early Sunday morning around the harbor!



Marybeth Kruppenacker, Christy Greeley and Carol Hughes exhibit for CRN at PAS.



Dr. Aniruddha Vidwans stopped by to check out the debut of CRN's new educational display.



Recordati Rare Disease representatives stop by the CRN booth.



Carol Huges, Dr. Alex Constantinescu, Marybeth Kruppenacker, and Dr. Rick Kaskel.



Kole Binger, medical student and sibling to Kelen Binger who lives with cystinosis, exhibited at PAS poster session and stopped by our booth to say hello!



# Rare Disease Week on Capitol Hill 2019

By Clinton Moore, President



**RARE**  
DISEASE WEEK  
ON CAPITOL HILL

Rare Disease Week on Capitol Hill each year always brings together a huge crowd of patients, parents, advocacy groups, health care providers, and industry. The week consists of a variety of meetings and activities for everyone to take part in. Some of these activities are focused on teaching you how to approach your representatives while others are more for social time to meet and create friendships with other rare disease patients or groups. Each year I am amazed at the number of people from all over the world that come to meet with their representatives and ask for the additions or changes they feel their disease community needs.

Then the day comes when we literally “Storm the Hill”. Hundreds of people head to the office buildings and that’s where they start making a difference.

Many of these people are nervous at first but then you see them later in the day and they are all smiles. I often hear them say that it was much easier than they thought it was going to be. These people leave the offices with a feeling that they have made a difference. And they have. They have had their voice heard. They have educated their state’s representatives on the disease that affects them.

This year I was joined by three others from our cystinosis community. Emily and Carol McClary and Aimee Adelman. Although we are all from different states and had to meet with different representatives, our mission was the same. Raise awareness and make changes.

In years past I have always been the only person to attend from Delaware.

Until this year. I was pleased to meet up with 5 others from Delaware that were attending for their first time. It was amazing to see the growth in just a year’s time. These others attendees and I still keep in touch today. These type of friendships last a lifetime.

When the week comes to an end the ask doesn’t come to an end. Follow up emails are sent and check-ins are done throughout the year. I already look forward to next year and hopefully will see even more new faces.

Check out this video summarizing the week’s events, sponsored by Rare Disease Legislative Advocates (RDLA):

<https://youtu.be/FVcWGzY7pm8>

# Recordati Rare Diseases Canada Inc. announces Health Canada approval of CYSTADROPS® (cysteamine ophthalmic solution) for cystinosis patients

March 4, 2019 | Business Wire | <http://bit.ly/2x0Mp2u>



TORONTO — Recordati Rare Diseases Canada Inc., a biopharmaceutical company providing orphan therapies for patients with rare diseases, today announced the approval of CYSTADROPS®, the first cysteamine solution approved by Health Canada for the treatment of corneal cystine crystal deposits in patients with cystinosis, from two years of age.

Cystinosis is an ultra-rare genetic metabolic disease that causes cystine – an amino acid – to accumulate in the body’s organs. It affects the kidneys, eyes, liver, muscles, pancreas, brain, and white blood cells, causing serious complications, including muscle wasting, growth delay, diabetes, hypothyroidism, and eventual kidney failure. The disease has an estimated incidence of 1 to 2 per 150,000 births, with up to 100 Canadians thought to be affected today.

“Recordati is excited to launch this newly-developed viscous eyedrop for cystinosis patients,” says Paul McCabe, Vice President and General Manager, Recordati Rare Diseases Canada. “Until now, in Canada, there was no approved cysteamine eye

drops solution for the management of ophthalmic symptoms in cystinosis patients.”

Ocular manifestations of cystinosis can cause significant long-term morbidity and develop into more serious ocular complications, including blindness. Left untreated, cystine crystal deposits form in the cornea, causing light sensitivity, eye pain, involuntary closing of the eyelids, and degeneration, resulting in partial or complete loss of vision.

“Unfortunately, current forms of cysteamine eyedrops require hourly application throughout the waking day, making sustained treatment difficult,” says Dr. Paul Goodyer, Professor of Pediatrics and Human Genetics, McGill University Health Centre, and a leading expert on therapeutic strategies for hereditary renal disease. “The recent arrival of CYSTADROPS® in Canada may represent a step forward. The drug is delivered in a viscous vehicle, thought to prolong exposure at the eye surface, and is administered only four times per day. Further studies are needed to understand how to optimize CYSTADROPS®’ use, but its stability at room temperature for one week and reduced dosing frequency could well improve eye health among Canadian cystinosis patients.”

CYSTADROPS® (cysteamine ophthalmic solution, DIN 02485605) is a cysteamine solution approved in Canada for the treatment of corneal cystine crystal deposits in patients with cystinosis. For more information, visit [www.recordatirarediseases.ca](http://www.recordatirarediseases.ca).

## About Recordati Rare Diseases Canada Inc.

Recordati Rare Diseases Canada Inc. is a biopharmaceutical company committed to providing often overlooked orphan therapies to underserved rare disease communities in Canada. Recordati Rare Diseases Canada Inc. is part of Recordati, an international pharmaceutical group committed to the research and development of new specialties with a focus on treatments for rare disease.

Recordati Rare Diseases Canada’s mission is to reduce the impact of ultra-rare and devastating diseases by providing urgently needed therapies. We work side-by-side with rare disease communities to increase awareness, improve diagnosis, and expand availability of treatments for people with rare diseases.

The company’s Canadian corporate headquarters are located in Toronto, with global headquarters located in Milan, Italy.

## Family Support

By Jen Wyman, Vice President of Family Support

Have you ever heard the phrase “pressure makes diamonds?” It comes from the fact that coal, which is dark and dirty, when under pressure for a long period of time, turns into diamonds, one of our most valued jewels. The same is true of the human being. When we put out muscles under regular stress, they grow, when our immune system faces challenges, it becomes stronger. And when we face challenges in life, and approach them with the belief that they can make us more psychologically resilient, that’s exactly what happens.

Our community has been under extreme pressure in the last few months, more than usual it seems. It has been a difficult time. Many families have been struggling with health issues, enduring hospitalizations, and suffering deaths. There isn’t much you can say to a family undergoing these stressors. There isn’t much they want to hear. But what I do know is that they want to know that people who understand are nearby...maybe not logistically, but within a phone call or a text or

a Facebook post away. It doesn’t replace the physical presence or a hug, but it works. It’s sufficient when there are many miles between families who suffer from a rare disease.

We are a couple months away from our summer conference. It’s an exceptional year for our community, when we can come together after many months of being apart. It is an incredible time of learning, but also a very special time where we can meet the people we call our cystinosis family, interact with the medical professionals that fight hard for our quality of life and ultimately a cure, and reunite with friends we have met on this journey.

Cystinosis is dark and dirty and every one it touches feels its pressure, but the diamonds-the most valued jewels-are all of the special people who make up the Cystinosis Research Network. The people who endure struggle, suffer and ultimately sparkle and shine in spite of it all.

In memory of those we have lost their battle with cystinosis.



*“We shall find peace.  
We shall hear angels,  
we shall see the  
sky sparkling with  
diamonds.”*

*— Anton Chekhov*



*Tim and Jen Wyman*



*Kacy Wyman*



*Son, Matt and Cameron*



*Son, Jack and Libby*

# INTERESTED IN RESEARCH?



Our goal is to understand how the brain processes and integrates sensory information in young adults diagnosed with Cystinosis. We will use EEG to observe the reaction of your brain while you listen to sounds and view images.

If you would like to participate in our study or to know more about it, please e-mail us at: [ana.alvesfrancisco@einstein.yu.edu](mailto:ana.alvesfrancisco@einstein.yu.edu)

**Participants are compensated \$15/hour**

**Help with travel costs available**

The Sheryl and Daniel R. Tishman Cognitive Neurophysiology Lab  
Albert Einstein College of Medicine, Van Etten, 1<sup>st</sup> Floor, C-Wing  
1225 Morris Park Avenue, Bronx, NY 10461

[www.cognitiveneurolab.com](http://www.cognitiveneurolab.com)



The Cognitive Neurophysiology Laboratory at the Albert Einstein College of Medicine in New York is currently recruiting individuals between the ages of 18 and 30 years old for a brain research study looking at sensory processing and executive functioning in Cystinosis. The study involves two days of testing: One day during which we will be recording brain activity using EEG; a second day dedicated to cognitive testing. Help with travel expenses is available.

# Research Update

By Christy Greeley, Vice President of Research



*The Greeley Family (from left to right): Dave, Alex, Christy, and Jack.*

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 \$300,000 and has funded over \$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.

## PROJECT UPDATES

Study of Neuronal Structure and Function Changes in Cystinosis – 1-year Progress Report

Krishnan Padmanabhan, PhD – co-PI,  
John J Foxe, PhD- co-PI

University of Rochester School of Medicine and Dentistry

Year Two Extension: \$110,000, Total Grant \$220,000

**1. Project Summary:** The aim of this proposal is to understand both the neurobiological underpinnings of cystinosis and the effects of cysteamine on neuronal structure and function using this Cystinosis Knockout (Ctns  $-/-$ ). By understanding disease processes at the mechanistic level, we aim to advance understanding of how cystinosis affects the structure of neuronal circuits, identify which cell types in the brain are most affected by the disease, and link molecular and cellular phenotypes with broader circuit level alterations in the activity of neuronal networks. We detail the major activities and significant results below and outline the plan for going forward.

**2. Significant Results:** Aim 1: The lab has successfully built out new multi-photon imaging system which allows us to complete the experiments proposed in aim 1 in the lab without relying on an imaging core. As many lysosomal storage diseases like Tay-Sachs and Niemann Pick Type C (NPC) present with structural abnormalities in neurons such as enhanced sprouting of dendrites early, followed by dendritic degradation later in life. Other structural features of the neuropathology in these lysosomal storage diseases include alterations in dendritic spine plasticity, and appear to be similar to changes in dendritic tree structure, spine size, and spine shape in diseases of intellectual disability such as Rett and Fragile X. Post-doctoral fellow

Max Mehlman has been trained in in vivo imaging in anesthetized control animals (to establish baseline measures of dendritic morphology) and to image spines over multiple time points. Representative examples of in vivo imaged dendrites from primary visual cortex (V1) and mouse somatosensory cortex (S1) are showing in figure 1. In year 2, these control measures will be compared to those acquired from the Ctns <sup>-/-</sup> mouse (see below).

### 3. Key outcomes and

**achievements:** Personnel: The excitement surrounding the electrophysiology and imaging research program has facilitated the recruitment of a research fellow and 2 rotation students who have advanced various portions of the grant. Post-doctoral Fellow Max Mehlman is working on the imaging portion of the grant. Two additional first year-rotation students Luke Shaw, and Mark Strossel will be continuing the electrophysiology work outlined in Aim 2 including recording from control and Ctns <sup>-/-</sup> mice.

Publications and presentations: Researchers and the lab supported in part by the Cystinosis Award presented a total of 3 poster presentations at the society for neuroscience meeting. Graduate Student Emily Warner was awarded a 2018 Trainee Professional Development Award (TPDA) from the Society for Neuroscience where she presented her work examining behavioral differences in habituations among mice used to model disease processes. The Cystinosis Research Foundation was acknowledged in all of these presentations and the work generated considerable excitement among neuroscientists.

### 4. Goals for the next reporting

**period:** We are deeply committed to research goals of this project, and the importance it has for studying neural mechanisms of cystinosis. However, we have had difficulty in obtaining the Ctns <sup>-/-</sup> mice to continue our work in year 2. To address this, we are proceeding along an alternate strategy. In coordination with the University of Rochester Transgenic core, we are generating two strains of Ctns knockout mice for use in this project, and which will make available to the research community through the Jackson Laboratory Mouse repository. Briefly, we outline the strategy for knockout of the Ctns gene. The first approach is a constitutive knockout and the second approach is a Cre induced knockout that will allow us to target specific cell types at various stages of development to isolate potential targets for intervention. We are coordinating with the director of the Mouse Genome Editing (MGE) Resource, Dr. Lin Gan (Professor, URMC) to generate these lines. We will generate 2 strains of transgenic Ctns mice by gene editing mouse models using CRISPR/Cas9 approach. We have already initiated a strategy for generating these mice with an expected timeline to first breeders between 3-6 months depending on the efficiency of the CRISPR targeting.

### Mechanisms Underlying Neurocognitive Changes in Cystinosis – Interim Progress Report Year 3

John Foxe, PhD Co-Principal Investigator, Sophie Molholm, PhD Co-Principal Investigator  
Departments of Neuroscience and Pediatrics, Albert Einstein College of Medicine,  
Montefiore Medical Center

Total Grant plus Travel Addendum:  
\$338,322

### PROGRESS REPORT

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.

**Scientific communications:** We presented analyses of the data at the Cystinosis Research Network Family Conference in Utah, at the Pediatrics Research Day and at the Lysosomal Rounds at the Albert Einstein College of Medicine (April 2018), and at the International Meeting of the Psychonomic Society in the Netherlands (May 2018). We have started preparing the data from two of the datasets for publication and are currently working on two manuscripts.

**Personnel Change:** Dr. Ana Francisco arrived mid-September 2016 and has taken over as the key recruitment and data collection and data analysis member of our team. Dr. Francisco is now familiarized with the paradigms and the extant data and is managing the IRB protocol.

**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. Though we have met recruitment targets (in the past two years, 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 believe that increasing our adult sample would be important to better understand the developmental path of the processes being investigated. Given the additional funds recently granted by the CRN for this purpose, we will soon start recruiting adult individuals to complete our sample (both diagnosed with cystinosis and neurotypical controls).

**Paradigm Development:** In addition

to collecting data for our originally proposed study (results detailed below), we added to our protocol the Delis-Kaplan Executive Function system (D-KEFS) battery (to assess key components of executive functions within verbal and spatial modalities) and two EEG studies to probe the brain processes underlying executive functions in Cystinosis. In brief, one provides a sensitive assay of the ability to withhold a prepotent response, and the other measures the cortical network engaged during task-switching. Additionally, we included an auditory sensory processing task. These are tasks that we have used extensively in our work to interrogate brain function in healthy adults, over the course of childhood development, and in clinical populations.

#### **Year-3 Interim Conclusions:**

Multisensory processing in Cystinosis seems to be largely intact and developing normally in the younger patients. Although the recently acquired datasets still need to be added to the analyses, the present findings are in contrast with other lysosomal disorders. Continued analyses of the electrophysiological data will search for changes in neural timing and circuit-level neural activations to both auditory and visual inputs, and to the combination thereof. As described above, we have added paradigms to assess higher-order cognitive processing in this population. Using behavioral measures and high-density EEG, we are investigating brain processes underlying executive function in Cystinosis. The results from the neuropsychological D-KEFS test suggested significant differences in executive functioning between Cystinosis and control groups. However, those differences were only observable in time, not in accuracy. This might be an indication that

individuals diagnosed with cystinosis do not lack executive function skills, but rather require more time than neurotypical peers to carry out such tasks. One implication of this overall slower processing is that individuals with Cystinosis may be interpersonally slower but not unable to engage. The EEG data recorded during the response inhibition task revealed differences between the groups, at least in children and adults. In these age groups, the difference between go and no-go trials was larger for the cystinosis groups. These behavioral and electrophysiological results have implications for greater understanding of executive functioning and perhaps interpersonal functioning in individuals with cystinosis. Further analyses, such as the focus on the trials that follow response withholding and error-related negativity in each of the groups, will be carried out and may add to the present results. Nevertheless, larger adolescent and adult groups are needed to determine if the behavioral and neural pattern differences that we observe in children generalize to the older cystinosis population more broadly. The results from the oddball duration task suggest the presence of an automatic pre-attentive processing impairment in children and adolescents diagnosed with cystinosis. The small adult samples does not let us draw any conclusions about the resolution of this potential impairment. Again, adding more adults to our cystinosis sample would be fundamental.

Future Plans:

1. We will submit two manuscripts (executive function and auditory processing) by the end of year 3.
2. In addition, we plan to begin analysing data from the task-switching task. The behavioral and EEG results

from this paradigm might inform the response inhibition task results and contribute to a better understanding of executive function in Cystinosis. We also plan to assess whether or not some individuals are more impaired than others in each of the paradigms and test the relationship between brain and clinical measures.

3. We will attend the Cystinosis Research Network Conference in Philadelphia this coming July (2019) and present the latest results from our studies.

4. We will recruit adults, increasing the age range studied, so that we can address important developmental questions.

### EDUCATIONAL RESOURCES

All of CRN's educational materials including brochures, guides and other publications have been updated and

are available on the CRN Website. They will also be available at the Conference in Philadelphia in July for attendees.

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](mailto:bryantjo@mail.cc.nih.gov)

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 Publications and Guides library at <https://www.cystinosis.org/support-resources/publications-guides/>



*Jack and Alex Greeley.*

## The Cystinosis Research Network, Inc. Financial Review

*By Jenni Sextone, Treasurer*

For the 3 months ended March 31, 2019:

### Revenues

The income for the three months ending March 31, 2019, was \$311,000 compared to \$40,000 in 2018 due to increased funds for the 2019 Family Conference in Philadelphia.

### Expenses

Total operating expenses of \$117,000 were higher than operating expenses for the same period during 2018 of \$33,000. Research grant expenditures in the first quarter of \$70,000 was the primary reason for the increase in expenses as compared to the same period in 2018. Excluding the grant

payments made in 2019, operating expenses were \$14,000 higher than the same reporting period in 2018 due to increased expenses related to the website.

CRN had net operating income of \$194,000 for the three months ending March 31, 2019. Continuous fundraising activities and generous corporate support continues to provide cash resources to increase patient advocacy and family support activities in 2019 and beyond to support the cystinosis community. Cash on hand at March 31, 2019 was \$422,000. Net change in cash for the first quarter 2019 was an increase of \$62,000.



*The Sextone Family.*

# Stand Up to Cystinosis

By Herberth Sigler, Vice President of Development

Cystinosis Research Network (CRN) wants to express our gratitude to Bill Brink for raising cystinosis awareness through his February 2019 fundraiser: Stand Up to Cystinosis. Bill mentioned to us his idea and CRN supported him. Bill is someone that has our admiration and represents an immense inspiration to my family. I believe CRN and its members know, cystinosis is part of our lives and is a daily reminder we are looking for a better and strong network, improving treatment constantly and ultimately a cure.

He wrote this to us, "We created this event to help raise money for cystinosis because this disease is something very near and dear to my heart. I have been living with this disease since I was 4-years old. One day I was talking to my friend and trainer, JD, the owner of the gym. I mentioned starting a fundraiser making and selling t-shirts, JD mentioned a weight-lifting event and here we are".

Today, Bill is an adult looking forward, standing up every day for himself, for the CRN community and the message to the public is: It is possible! It is



doable! Personally, I can say looking at these pictures there are few words to express the complex and mixed feelings in my heart. Bill, please receive in the name of my family and CRN our admiration for what you have done!

Bill wants to give thanks to friends, he wrote, "to JD and Gaylynn Marcum without them and the use of Florence Strength and Conditioning none of this would have been possible. I would also like to thank all the lifters who participated, friends, family and spectators for your donations". CRN expresses thanks to those that helped him in every detail for this weight-lifting fundraiser. JD, we understand you had some difficult moments at the

personal level during the planning for this event. Please accept our profound thanks not only for standing next to Bill but also for your service for this country in your military career. Our prayers will be with you.

Feel free to contact me by email to [hsigler@cystinosis.org](mailto:hsigler@cystinosis.org) and we will be glad to support your initiative.

*"If you think you're too small to make a difference, try sleeping with a mosquito in the room."*

— Dalia Lama



# DNACheckup has Partnered with Dante Labs to Support Families Affected by Nephropathic Cystinosis

Dante Labs has reduced their prices for whole genome DNA testing (WGS) for patients with nephropathic cystinosis to \$299 (regularly \$599). The report provided by Dante Labs allows family members to test for the same genetic mutation (carrier testing) through DNACheckup for \$150 per family member.

As a carrier you might pass the mutation to your children (50% chance). Children of carrier parents (when both parents are carriers) have a 25% chance of inheriting two mutated genes and developing the disease. It is important to be informed to make timely decisions.

A nonprofit 501(c)(3) organization, DNACheckup provides safe, low-cost, doctor-prescribed genetic testing for families affected by recessive genetic disorders.



*“ We were able to have our family tested to see which gene my husband and I carried. We also had our girls tested to see if they were carriers. It also confirmed our son’s mutations. ”*

*- Jodi G.*

*“ DNACheckup was amazing in helping with family planning. Easy to use, great customer service, quick results! ”*

*- Sandy R.*



## CREATE ACCOUNT & ORDER

Please create an account for each individual participant. Once your account is approved, you will be able to order the test and your cheek swab kit will be shipped.



## COLLECT SAMPLE AT HOME

Once you get your kit it's just a quick swiige inside your mouth to collect the sample.



## SHIP YOUR SAMPLE BACK

Send collected sample back to us for testing. A prepaid return envelope is provided with the kit.



## RESULTS AVAILABLE ONLINE

Once we get your sample, we'll handle the rest and let you know when your results are available in your online account.

**Please note:** most donations received via Facebook fundraisers are filtered through and categorized as contributions via Network for Good. Our heartfelt gratitude goes out to all who have reached into their pockets to support the cystinosis community.

## 2018 Donor Honor Roll

### \$100,000+

Horizon Pharma USA, Inc.

### \$50,000-\$100,000

Leadiant Biosciences, Inc.

### \$25,000-\$50,000

Lincolnshire Sports Association

### \$25,000-\$50,000

Arnold, Norman J. & Gerry Sue

Network for Good

### \$2,500-\$5,000

Carmichael, Scott and Tia

Schleuder, Don L.

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**CYSTINOSIS**  
**RESEARCH NETWORK**

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**Editor** Christy Greeley

Cystinosis is a rare, genetic, metabolic disease that causes an amino acid, cystine, to accumulate in various organs of the body, including the kidneys, eyes, liver, muscles, pancreas, brain and white blood cells. Without specific treatment, children with cystinosis develop end stage kidney failure at approximately age nine. 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.