

THE CYSTINOSIS Advocate

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2019 Family Conference Philadelphia: Ring in The Future

Individuals, families, world expert clinicians, and industry partners came together this summer to help our cystinosis community "Ring in the Future". The city of Philadelphia welcomed:

80 people living with cystinosis | **9** different countries | **275** overall attendees

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The President's Letter

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Maya Doyle, MSW, Ph.D., LCSW-R

The Thanksgiving season seems to be the perfect time to be drafting this letter. The past 6 months have been a very challenging and worrisome time. From battling health challenges to researching insurance options to looking for specialists, needless to say... its been busy and frustrating.

Thankfully I have had a great number of individuals in this community reach out to offer advice, share their experiences, and give suggestions on our next steps. As these may seem like very small efforts to those individuals, they have been huge in our decision making and search for answers. I can't thank this community enough for their outreach and support during this time. Not only have we felt supported but it also led us to the changes that are upcoming in our healthcare.

Nearly everyday, I see members of this community doing this exact same thing with each other. It's so easy to see that no one in this community is ever alone. It's amazing to watch how everyone just jumps in and offers the assistance that they can when they all have their own worries and fears going on. Selfless acts everyday. It's just beautiful.

So for this holiday season, I am thankful for this community of individuals that constantly give and offer what they can. I am thankful for all the ones that reached out and helped my family in our time of need. Many of you said you wished you could do more... but in reality you already did so much. I am forever grateful. Happy Holidays.

Clinton Moore



Chandler and Clinton Moore.



The Moore Family enjoying dinner at the Philly conference.

2019 Family Conference Philadelphia, continued

Families experienced a weekend filled with critical research updates from clinicians and international cystinosis organizations, opportunities for study participation, interaction with health care professionals, and a chance to meet old friends and make new connections. For those unable to attend, the live streamed sessions are now accessible on the CRN YouTube channel.

We were honored to provide unique experiences throughout the conference, some of which we have highlighted below. A special thank you to all who attended, those who contributed to making this event a success, and each and every person who champions for improved treatments, improved quality of life, and a cure for cystinosis.

#CRNPhilly Hope Fence Lock Project

After receiving a lock, heartfelt sentiments were written on the back of each. One by one the locks clicked into place to form a “C”. This was our moment to reflect on friendships, hope, storytelling, and the disease that brought us all together.

Walk in My Shoes – A Cystinosis Documentary

The trailer for the first-ever cystinosis documentary debuted at this year’s CRN Family Conference. You too can walk with the Moore Family as they recount their cystinosis journey and the steps leading to their physically and mentally challenging Cystinosis Awareness Day fundraiser.

Facing a Bright Future: Cystinosis Comic Book Series

Author, illustrator, and adult living with cystinosis, Kevin McCalla revealed the third and final installment of his original cystinosis comic book series. “Facing a Bright Future” ships free internationally. Sign up for your free copy.

Research and Treatment Opportunities

Dr. Larry Greenbaum and his research staff recruited individuals to participate in his Grip Strength Study, which also took place at the CRN Conference in 2015. He hopes to determine risk factors for decreased grip strength and to determine the severity of the problem in patients with cystinosis. Describing the problem will hopefully lead to studies of interventions to slow or reverse muscle strength.

Dr. Bruce Barshop provided opportunities for convenient White Blood Cell testing at the conference.

Adults Leadership Advisory Board (ALAB)

Adults living with cystinosis are a main focus of CRN. To that end ALAB was launched, a CRN initiative comprised of adults living with cystinosis who will develop further programming and opportunity for adults affected by cystinosis. ALAB members participated in a variety of ways during the conference, including hosting the family dinner on Friday evening, participating in panels focused on the issues facing adults, participating in an in-person ALAB board meeting, facilitating a breakout session for teens and adults to discuss ideas and resources for the teen and adult community, and finally serving as panelists to wrap up the event, answering questions for those in attendance on their experience living with cystinosis. We are proud of their growth as the future leaders of the cystinosis community and their contributions to this conference.



Hope Fence Lock Project.



Kevin McCalla, cystinosis comic book author and illustrator.



Grip Strength Study.

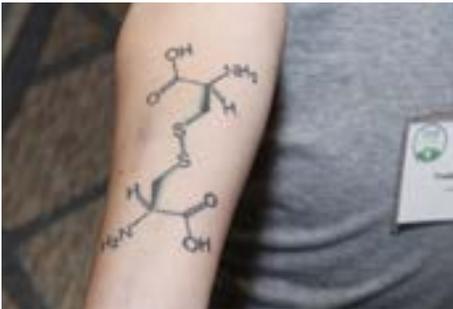


Dancing the night away.



Adults with Cystinosis panel.

2019 Family Conference Philadelphia: Photo Gallery



2019 Family Conference Philadelphia: Photo Gallery





Adult Leadership Advisory Board: A Look Into Our Future

By Cheryl Simoens, ALAB Chairperson



ALAB members met up at the 2019 Family Conference in Philadelphia.



ALAB Chairperson, Cheryl Simoens.

I am thrilled to be the newly appointed Chairperson for the Adult Leadership Advisory Board (ALAB) with the Cystinosis Research Network (CRN). Growing up with cystinosis was challenging at best, as I did not have the support, resources or comradery I have today in my life. I often felt isolated and asked questions like, "Why is this happening to me?". However, I learned how to be a fighter and do not let the tough times hold me down, like so many others in this community. Being a member on this Board will allow me to be a part of a team that is whole heartedly dedicated to providing meaningful services, programming and opportunities to engage the whole cystinosis community. I am devoted to supporting, listening and learning from individuals around me and helping to instill the importance of resiliency and being a fighter, as we all have a story and together, we are stronger.

The passion and dedication I have witnessed while working with both the ALAB and CRN has inspired me and ignited a new level of passion. I look forward to working with the ALAB to create and implement services to support the entire cystinosis community. With immense eagerness, creativity and commitment, we can achieve a great number of projects

benefiting generations to come and share stories of strength, hope and inspiration.

It is with great excitement to be announcing two additional leadership roles with the ALAB, that of Vice Chairperson and Secretary. Please join me in welcoming Sara Healey as Vice Chairperson and Karen Gledhill as Secretary. I am proud to be a part of this incredible committee, working with such brilliant individuals and ready to tackle the future on this board!

Our collective team has been busy working on three new projects launching soon. Each initiative was created by an ALAB member to support our mission: to share our stories and strength to educate, motivate and empower the entire cystinosis community.

Below are further details and descriptions on these projects:

CystinosisTEENS: Instagram Account

A private/closed Instagram account focused on tackling issues, spotlighting individuals, and providing a safe forum for teens with cystinosis to connect.

Cystinosis Sessions: Video Conferencing Program

Providing a face-to-face (video)



ALAB Vice-Chair, Sara Healey.



ALAB Secretary, Karen Gledhill.

platform to share experiences and educate cystinosis patients, parents, caregivers, and healthcare professionals in a knowledgeable and comfortable environment.

Cystinosis Rare: A Journey into the Unknown (Podcast)

Quarterly podcasts, hosted by individuals living with cystinosis, delivering community and provider input, highlighting topics such as mental health, cystinosis success stories, teen to adult transition, and education system challenges. Designed to support, educate and empower.



I would like to take this moment to remember a wonderful, enthusiastic and loving member of the ALAB who we lost over the fall, Laura McGinnis. Laura will remain an honorary member on the ALAB for her efforts, talent and positivity during her time on the ALAB. She will be missed by so many and forever remain in the hearts of the cystinosis community.

American Society of Nephrology Conference: Kidney Week 2019

By Heidi Hughes



Heidi Hughes shares her first-hand experiences with visiting professionals.

Carol and Heidi Hughes, Clinton Moore and Gail Potts were privileged to attend the American Society of Nephrology (ASN) Conference in Washington, DC from November 7-9. This was a very well attended conference with 13,000 participants. The Cystinosis Research Network exhibited all three days with literature, brochures for professionals explaining the treatment and transition of cystinosis patients. There was interest in the transition phase by several physicians who stopped by our booth.

We spoke with a physician who started a support network for his patients in India and was looking for information about our advocacy organization. We were able to provide him with information about CRN.

We had a physician consult with us on how to deal with a patient who was non-compliant in following up

his cystinosis with other physician specialists. We suggested some of the mentoring available through [ALAB \(Adult Leadership Advisory Board\)](#).

We had the opportunity to network with Horizon while there also.

I believe we helped to inform physicians and other professionals along the road to better understanding of cystinosis. Having Heidi, an adult living with cystinosis, there to answer personal questions was wonderful. Many stopped to talk with her and she was able to share first hand her story.

As I descended down the stairs for my first American Society of Nephrology(ASN) event, I was overwhelmed (in a good way). Pharmaceutical companies, doctors and Kidney Week enthusiasts oh my! This was such a wonderful opportunity and privilege to represent Cystinosis Research Network and the Adult Leadership

Advisory Board (ALAB) at an event where I was the face and voice for my fellow warriors.

Clinton, Gail, my mom (Carol) and I all manned the CRN exhibit. We had over 50 professionals from all levels and countries visit our corner booth during this action packed weekend. Throughout these encounters, we shared stories, advice, and tons of literature with each interaction ending in a hug, hope for the future of Cystinosis and enlightenment for all. I shared about medication (in) tolerance and struggles throughout my childhood, trials and tribulations of life with Cystinosis, and a perspective that many medical professionals tend to overlook and not have the opportunity to experience outside of their clinics. We all served as the visual aide for both struggle and success with this disease for both patients and caregivers alike. I truly felt that we all left a strong impact of the importance of community and CRN's role in the success and research as well as

educating and installing knowledge and hope at ASN.

While exploring the exhausting amount of booths and elaborate showcases by the pharmaceutical companies, I spoke to multiple companies from Horizon and Leadiant to the National Kidney Foundation and the American Kidney Fund to both gain and share knowledge of our roles in the treatment and involvement with our little Cystinosis community. This truly brought a light to the importance of our involvement in medical conventions and events to spread awareness and gain support. I took this chance to share with them our experiences with the drug interactions all the way to the ordering process and the many pains and headaches in between.

This was a privilege and opportunity. My hope is that everyone we came in contact with took away the hope and education CRN provides to doctors, families, patients and caregivers around the world. Our impact goes beyond the borders of our country, posts on social media, and doctor's offices. Together, we have the ability to share our stories and forever impact the medical community by attending these conferences and bringing our perspective and strength.

Thank you CRN for this amazing opportunity!

Heidi Hughes



Brent Lechner, MD and Heidi Hughes.



Muscle issue conversation with Dr. Grimm.



From left to right: Alhamad Tarek, MD, Umar Farooq, MD, Heidi Hughes, Clinton Moore, and Gail Potts.



Mingling with industry partners.



Heidi Hughes with Oulimata Grossman, MD.

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Ireland Update: Worldwide Cystinosis Community Advisory Board (CAB)

Cystinosis Ireland is proud to be hosting Cystinosis Network Europe in their work on the International Cystinosis Conference 2020 and the Worldwide Cystinosis Community Advisory Board (CAB).

The cystinosis CAB has developed and is supported through the EuroCAB programme, a project of EURORDIS, the European Rare Disorder Organisation.

The CAB's objective is to improve patient access to novel therapies and treatments. This is achieved by engaging with clinical trial sponsors at the earliest stages of their research processes. The CAB also works with pharmaceutical companies on topics like educational materials and other appropriate topics. As well as meeting with industry sponsors, the Board engages with early-stage researchers as part of PPI - Public and Patient Involvement in research. We are also keen to work with physicians/clinicians in any way that might be helpful, in particular around research, drafting clinical guidelines, etc.

The CAB has a wide remit to engage on behalf of the cystinosis patient community. The members of the CAB are parents of children with cystinosis and adults who themselves have cystinosis. Members of the CAB are nominated by the cystinosis patient organisation(s) in their own countries. They are educated and trained in clinical trials, drug development and research and provide the patient voice and patient expertise.

The first meeting of the Worldwide Cystinosis CAB was held in Dublin, Ireland at the end of October and early November and included representatives from Ireland, Northern Ireland, Spain, France, the Netherlands & Belgium, Germany, the United Kingdom, and the United



From left to right: Will Newman, Christy Greeley, Anne Marie O'Dowd, Markus Schmitt, Irati Vilarino, Marjolein Bos, Ruth Davis, Fons Sondag, Lilli Sanz, Nicole Patin Paybaud, James Ennis, Sonke Oetjen.

States. These patient advocates reviewed research proposals and met with future sponsors of clinical trials to discuss their plans and provide expert input. The meetings took place over four days with a heavy workload and the group found the various meetings and discussions informative, engaging and beneficial.

The Worldwide Cystinosis CAB plans to meet again in July 2020, in advance of the International Cystinosis Conference.

The International Cystinosis Conference 2020 will take place in Dublin, Ireland from 9-11 July. We are delighted that Prof Elena Levchenko has agreed to be our international chair, and Dr Atif Awan our local chair for the meeting. The agenda is currently being finalized and will be circulated when confirmed. We are also planning a full childcare programme and specific agendas and social programs for adolescents and young adults living with cystinosis.

You can find out more on www.cystinosis-europe.eu or email denise.dunne@cystinosis-europe.eu.

Seedcorn Funding

Cystinosis Ireland continues to offer Seedcorn funding of up to €10,000 over 6 months to researchers looking to generate solid preliminary data which would lead to a larger, sustainable, longer-term application for funding. The scheme is open to researchers new to the field of cystinosis, and welcomes projects in blue-sky, high-risk areas. More details including Cystinosis Ireland's priority research areas can be found on www.cystinosis.ie/research.

Closing dates for applications are 5 March 2020 and 31 July 2020. All queries should be directed to research@cystinosis.ie



Cystinosis Network Europe International Conference

Bringing families & research together



Save the Date!
Dublin, Ireland
10 - 12 July 2020

Mexico Update: Fifty Seven Years with Cystinosis

By Gerardo Mendoza Valle



My name is Gerardo Mendoza Valle. I'm Mexican. I am 57-years-old and I was born with cystinosis. I am alive thanks to three kidney transplants of cadaveric origin (1977, 1986, 2007). Since 2007 I take Cystagon and I hope I will not need a transplant again. Currently I face other diseases: hepatitis C, hypothyroidism, hyperparathyroidism, pneumocystis, cancer and loss of vision in the left eye due to corneal rupture. My only sister was also born with cystinosis, died at 20 years of age in 1984.

Living with cystinosis has not been easy, especially in the times when kidney transplants were not common, when the hemodialysis treatment took seven hours per session, when there was no erythropoietin or Cystagon and when in Mexico nobody knew cystinosis. However, I am convinced that what helped me the most was that my parents never treated me as sick. I had a very happy childhood, my school performance was normal, I finished university and I did postgraduate studies. I got married in 1996.

Living with cystinosis gives us the



opportunity to value what we have and make the most of it, especially in times of stable health. I know 33 countries around the world, I have practiced various sports such as hang gliding and kayak river descents, I take great care of my health with a vegetarian diet for 33 years, I practice hiking, yoga and meditation.

However, what really makes me happy and grateful is to help others, especially people who suffer the same as me. In 2002 together with Víctor Gómez, patient with cystinosis and a few more families, we created the Asociación Mexicana de Cistinosis A.C. (Cystinosis Organization Mexico). Since then we have helped other people with cystinosis and parents of children with this disease.

In addition to AMECIAC I co-

founded three other foundations: the Asociación Mexicana Pro Renal Transplant A.C.; The Center of Mexican Corneas; and the Holbox Island Foundation. In 2001, I was invited to be the General Director of the National Transplant Foundation (FUNAT) where I had the opportunity to transplant 7,200 Mexicans in ten years, my greatest achievement.

Living with cystinosis is a challenge like many others. The support of family and friends is fundamental, but it depends on us to determine the attitude we take in the face of the adversities that this disease presents to us. And there will always be more reasons to thank than reasons to complain about.

Thank you.

Gerardo Mendoza Valle

Mexico City

gerardomendozavalle@gmail.com

Education and Awareness Committee Update

By Marybeth Kruppenacker, Vice President of Education and Awareness



Marybeth and her family.



Marybeth's grandchildren.

As I write this article I feel humbled that I have been asked to take on the role of Vice President Education and Awareness...again! I truly am honored to take on the responsibility that was bestowed on me when CRN was formed over 20 years ago. I suppose that is good! It actually is very good as I look around this organization and know who we are and just what we have become as a rare disease advocacy group...and it is extraordinary. CRN has grown in so many ways as we all work together and continue with our mission of new and improved treatments and an eventual cure. CRN is recognized as a "model" advocacy organization and I believe one that has been looked to for guidance in many ways around the world. I am so proud of that!

CRN continues to be "at the table" and not only attend but often times asked to participate in meetings and symposiums around the world. We still exhibit at two professional meetings each and every year, the pediatric and adult nephrology meetings, as well as, the Global Gene's meeting, and Rare Disease Day to name a

few. We are proud to represent and interact with medical professionals and educate them and passing out our literature to them to share with families as they return to their practices. Not only do we meet and speak to doctors first hand, but they are always complimentary and impressed that we have the ability to attend these meetings.

I am reflective also as September marked the 30th Anniversary of my now almost 34-year-old daughter's diagnosis! Little did I realize that 30 years ago we would be looking at a future of unlimited possibilities. Laura is the first generation to take cystagon (she actually was in Dr. Gahl's study pre-Cystagon) she is a 20-year kidney transplant survivor as well as a two year breast cancer survivor. I am so proud of who she has become despite cystinosis, but that reality does have a way of creeping in there and those opportunities are not always without struggles. I know that first hand as the mom and caretaker of a young adult with cystinosis, it is also all about how you handle every single

day but also how you take care of yourself and the opportunities each challenge presents. I believe that is the outlook that all of us at CRN have when it comes to opportunity. CRN has been blessed in so many ways and I would like to see us continue to work together towards a productive and bright future for everyone who is impacted by this rare disease.

So I look at this chance to serve again as an opportunity to continue what we started many years ago and I am truly honored to be a part of such a special organization and group of individuals who I consider to be my friends. We will continue to do great things and seize every opportunity.

Marybeth Kruppenacker

Attend Kidney Week 2020
October 20-25, 2020



Denver, CO
Colorado Convention Center

FAMILY STORIES: Following Your Intuition: Our Road to Diagnosis

By Anna Pruitt



Abel with an NG tube, December 2015.



The Pruitt Family.

November 18, 2015

is when we first were given the news that our oldest son, Abel, could possibly have cystinosis. We were only given a short transactional response from a renal doctor, in a prestigious hospital that lasted only seconds before she left the room to see another patient. Her apathetic response that our son could have cystinosis forced us to Google what cystinosis meant. Google informed us of how our lives were going to be changed forever.

In the dark depths of our undiagnosed moments that lead to our confirmed diagnosis of cystinosis, I was beginning to be molded into the best advocate for my children. The biggest lesson I've learned was to always trust my motherly instinct, as I believed this saved my son's life, and to have access to the resources that are out there. The resources that are provided on the Cystinosis Research Network web page was and still is a crucial asset for our journey, living with cystinosis.

September 2015

is when our son started to become

violently ill. He constantly threw up and cried for days on end. We went to several doctors and emergency rooms to try to figure out why our son was so sick. The doctor's constant response was that he contracted a virus and/or had a milk allergy. The doctors reassured us that it would pass in a couple of weeks. After a week, he never stopped vomiting and he was beginning to waste away right before our eyes because he couldn't keep anything down. That's when I knew as a mother and nurse that something was terribly wrong with him. My gut told me that something wasn't right and that I could not rest until I had it figured out. At one point in our journey, my motherly instinct turned into anger, rage and determination. This fueled my instinctive behavior to find answers and let my voice be heard. I became the squeaky wheel. I was the mom that called the doctor's office every hour to talk to the next doctor that would believe me, that something was wrong with my son and to order more tests. I became a person that I didn't want to be, but I knew I had to be, in order for these

doctors to listen to me and to take my concerns seriously.

December 7, 2015

was our confirmed diagnosis date of cystinosis. We were bombarded with a room filled with doctors that made countless of promises to take better care of our son. My persistent behavior became known to the hospital and doctors that finally gave us the results of Abel's cystine test. This particular hospital did not agree with my behavior and they tried to make me feel inadequate for doing so. No parent should feel inadequate when they are fighting for their child's life. We felt that these doctors measured their quality of care by their ego rather than compassion for what we were going through. It took a lot of instinctual fighting and factual clinical data to get our son properly cared for at this particular hospital and that took a toll on us.

After receiving the news that changed our lives, this hospital yet again failed us. The promise to order cystine-depleting therapy (Cystagon) and to schedule his G-tube surgery was left and forgotten by the staff and by

the renal doctor. After many phone calls and complaints, it took several days to get Abel scheduled for his G-tube surgery. By this time Abel was progressively getting worse.

The day of his outpatient G-tube surgery and just 2 hours before going into the hospital to be checked in, my motherly instinct kicked in again. I woke up with a knot in my throat and my gut telling me, "Do not go back to that hospital." I got onto the Cystinosis Research Network web page. There I found doctors that have treated patients with cystinosis and their affiliated hospitals. I scrolled through the list, tediously trying to find a hospital that could help. I found three doctors and two hospitals in St. Louis Missouri. I called both hospitals and every renal doctor that was listed. I remember calling and saying, "My name is Anna and my son was just



diagnosed with cystinosis, can you help me?" It was the 3rd phone call I made and a nurse named Grace had answered the phone. She knew what cystinosis was and after a brief conversation she stated, "Get in your car and get here ASAP! Abel needs to be admitted and needs to be stable before any procedure. He is not well." That phone call changed our course in treatment and we drove seven hours directly to that hospital not knowing how long we were going to stay. We only packed a couple of days' worth of clothes and we got on the road after loading the car in 30 minute. I remember the uneasy feeling - was I making the right choice in driving so far away to seek medical treatment? But as I look back, I know that my instinct said, "GO!" And so we went and that was the best decision we made for our family.

Without the resources of CRN and without following my instincts I cannot tell you if Abel would be here today, healthy as he is. We were immediately admitted and we stayed 15 days to get him stable enough for his G-tube surgery and to get all his medications just right. We came home with a PICC line, boxes full of medications, standing lab orders to draw labs

every two weeks and G-tube supplies. We had a team of compassionate nurses and doctors on our side. We had finally received the care that we desperately searched for and that couldn't have been done if we stayed on the same path.

October 2019

We have 2 handsome boys, both with cystinosis.

We are expecting our third child, Adaline, in January 2020. Paul was diagnosed six months after Abel and we started him on Cystagon one month after birth. Both boys are happy and healthy to this day. They both enjoy preschool and they are thriving. We moved to Colorado Springs, Colorado in August of 2018 to find even better medical treatment and for cooler weather. I continue to listen to my motherly instinct and our battles have been fewer since then. I cannot thank the CRN enough for the support and the website that we still continue to use. We encourage other families to have the courage to follow your instincts and to know you will always have the support from our cystinosis community.

The Pruitt Family



FAMILY STORIES: The Bartkowskis

By Jen and Ross Bartkowski



Ethan, 10 years old, celebrating Independence Day.



Ethan, 22 months old, showing off his tattoos.

Ethan is a caring, thoughtful, respectful, strong-willed, funny and smart 10 year old boy. He enjoys watching movies (Adam Sandler is his favorite actor), playing board games, watching YouTube videos, and he just recently found a love for basketball. He has overcome many obstacles to get to the fun-loving and energetic boy he is today and we have learned so many unforgettable lessons along the way.

Ethan was born on December 6, 2008. He was our first child and we couldn't have been happier. He was stubborn from the get-go and after 27 hours of labor, was delivered via C-section. We spent a couple days in the hospital before returning home and for first time parents, we thought we had everything prepared for our return with our new bundle of joy. Wow, were we wrong!

Our first night at home, Ethan would not stop crying and after about three hours we had tried everything. We reached out to everyone we knew and after the feeling of failure set in; we called the local emergency room and asked for help. They simply told us that they could not offer advice over

the phone and that we should bring him in and they would take a look. We loaded everything up, as new parents we NEEDED everything, and we finally got in the car. About 10 minutes down the road, Ethan finally cried himself to sleep. It was bittersweet... silence... thoughts running through our heads... do we still need to take him in?... will he start crying again when we stop the car and try to move him?... should we just take turns sleeping in the passenger seat as the other one drives and keeps the car moving?... what should we do? We drove around for another hour and decided to come home because it was almost time for Ethan to eat. When we got home he was content with eating and then went back to sleep. We weren't going to let this unexplained bad night ruin what we expected our new family life to be.

In the first three months we were admitted to the hospital twice. The first time was RSV and pneumonia and then a couple weeks later he got the Rotavirus. It just seemed like Ethan had really bad luck and we couldn't catch a break. We knew that being first time parents, we were going to encounter things like this and we just

needed to roll with the punches. We have a very vivid memory while in the hospital when a doctor came in to see Ethan and we explained that he had been crying and vomiting a lot, the doctor explained to us that it is called "spit up". This was obviously a very frustrating response to hear and we tried to explain that we knew what spit up was and this was actually projectile vomiting and he did it several times a day. The doctor just dismissed us and our concerns to being "first time parents", but we did get some validation when the doctor started to walk away and Ethan shot vomit across the room all over the doctor. We apologized up and down, but we were laughing on the inside.

The next couple weeks went by and Ethan ate/drank a ton. He cried a lot... the only thing that would soothe him was being fed or drinking water from his bottle. However, he would drink and then vomit, and continue that cycle over and over again all day. We tried switching from breast milk to formula and when that didn't work we went a formula hunt. We tried five different kinds and nothing worked, he would drink any of them... flavor didn't matter... but he would throw them all up within 15 minutes after eating. It was not uncommon for Ethan to vomit 25-30 times a day, and to specify... I mean vomit... not spit up... and between feeding and vomiting episodes he would just cry.

We went and saw the pediatrician several times, but since Ethan was growing and doing great on the weight/height charts, he wasn't concerned. He tried to offer us suggestions on feeding methods, formula choices, types of milk, use

of cereal with the milk, feeding less but more often, etc., but in the end nothing worked. Since he was growing he was getting what he needed and there was not cause for worry.

By Ethan's one year doctor visit, we continued to experience the same issues, nonstop crying when he wasn't being fed and vomiting all of the time, however now we had the added fun of baby food color in the vomit. He was no longer on the high end of the height/weight charts, however he was still at normal ranges so there little cause for concern by the doctors. We had also been through several daycare providers, family members and friends to watch Ethan during the day while we were at work because the constant crying and vomiting was "trying" to say the least.

It wasn't until a visit around 15 months of age that the doctor began to get concerned. We had explained that Ethan would no longer eat food and only wanted milk. Ethan was not gaining weight and actually had lost weight in the previous three months. The doctor recommended we go have tests taken at the hospital, because he had a concern that there was something wrong with his esophagus.

Within the next week, we went into the hospital and had several swallowing tests done, diagnosis was "EE" (Eosinophilic Esophagitis). Basically, a fancy name for "allergic esophagus", his esophagus would get inflamed when he would eat because he had more eosinophils than the average person. They prescribed him a steroid inhaler and pills that would need to be crushed and mixed with sugar that he would need to swallow before eating to reduce inflammation. We did not notice a change at all with the vomiting, but we were hopeful we finally had our answer and things

would get better.

A month went by and we went back for our follow up visit. We had tried everything, and he still wouldn't eat. He was losing weight and had no energy, we were at a loss. We did everything we could, but he was just miserable. The doctor diagnosed him with failure to thrive at that point and I will never forget the words he said "we will give you a couple more days to try and get him to eat but if you can't he will need to have a feeding tube placed". These words still give me chills thinking about them. We were devastated, we failed as parents, how could we not get our child to eat, a feeding tube...isn't that only for people that are dying? We had so many thoughts and concerns. We left the doctor appointment numb.

Two days went by and we knew it was inevitable. Our little boy was not going to eat by mouth and there was nothing we could do. We called the doctor and scheduled the hospital visit to have the feeding tube placed. We thought this was the end of the world... unfortunately without us knowing...the world wind had just begun.

We got up in the morning early to take Ethan to the hospital and have his feeding tube placed. Before the surgery could be performed they do routine blood work and according to our surgical team, this was all standard. Once the results came back, they would move forward with the procedure and it was expected to be short and we would have our baby back in our arms. Unfortunately, that is not how it played out at all. Within 15 minutes we had our results and Ethan was being rushed to the ICU. His potassium levels were at 1.5 (normal child range is above 3.5) and the rest of his levels were a mess. They explained (very fast) that they



Ethan, 4 years old.

were worried he would go into cardiac arrest at any time and he needed to be moved to the ICU. They luckily already had an IV hooked up for the surgery, so as they were moving him to the ICU the nurse was hanging a new bag with everything needed to get his electrolytes stabilized.

Days went by in the ICU, it was all a blur. Sleepless nights of walking the halls to soothe him and reduce the crying so other children could sleep, trying to Google possible reasons for Ethan's issues and most of all...just trying to make sure our baby was OK. The medical staff focused specifically on stabilizing him. They did a great job and watched him day and night making sure he had everything he needed. They started him on TPN (nutrition through his veins) so he could start to get some nutrition and he would hopefully not vomit as much. He continued to drink water out of a sippy cup to soothe himself, because that was the only thing that made him feel better. I can remember so many specialists, doctors, interns, PAs, RNs, etc., coming in and giving Ethan a "once over" to try and determine what was going on with our child. His

previous diagnosis of Eosinophilic Esophagitis would not cause his electrolytes to be so far out of balance but he didn't have anything else that stood out to any of these doctors as "abnormal".

It wasn't until day five of the ICU that we had a very special doctor, he was a genetics doctor, come in and talk to us. He asked us some very "weird" questions like "can I see your hands?" and then he would say "no that is not it". After about four random questions, he was onto something. He asked "does Ethan like to play in the water? Does he act like the sun bothers him? Is Ethan always thirsty?" After we answered yes to a bunch of his questions, his response was "OK, thank you...I am going to have a specialist come in tomorrow and take a look at Ethan's eyes, I think I may know what is going on...but I do not want to alarm you so we will just wait until tomorrow". UM, NO... We could not just let this man leave the room that had an idea on what could be wrong with our son, when nobody else had any ideas...we had just spent five days in this room and needed something...anything. We begged him to please tell us what he thought. He gave in, but explained that he didn't want us looking online on what "cystinosis" was until we had confirmation from the eye doctor and the blood tests (needed to fully confirm). Needless to say, he walked out of the room and we grabbed our laptop and started uncontrollably reading and trying to understand what "could be wrong" with our baby.

The next day was a whirlwind of emotions. The eye specialist came in, confirmed that she could see crystals in Ethan's eyes, and therefore the only other test that was needed was the blood test. The blood test needed to be sent to a lab in California and



Ethan (10) with brother Jackson (7) at the Cystinosis Research Network Conference in Philadelphia, summer 2019.

therefore it would take some time to get back. However, the doctor confirmed that there were no other known diseases that would cause crystals to form on a child's eyes and therefore he was fairly certain Ethan had "cystinosis".

Days turned into weeks and weeks turned into almost two months. We were transferred out of the ICU and put on the GI floor for monitoring. During this time we tried everything to get him the nourishment he needed, help control the vomiting, and get his electrolytes stable. Unfortunately, after many sleepless nights and trying to determine the best solution for Ethan, we were running out of options and his continued vomiting would not allow his electrolytes to stabilize. The decision was made to have a permanent feeding tube placed, this would not only help with nourishment but it would also help us be able to administer the meds he needed around the clock. It was also decided to place a central line into his heart to provide him with the necessary electrolyte supplements and hydration. These decisions were not made easily, but we felt it was the

best option for Ethan at the time.

After we got out of the hospital, we had to adjust to our "new normal". Ethan was connected to an IV pole for 20 hours a day and had to be monitored 24/7. We required at home nursing care while we were at work to administer meds, operate the central line and run the feeding tube. Welcoming people that you don't know into your home to take care of your very sick child was definitely one of the harder adjustments we had to make, but we were so lucky to have some great caregivers that became our second family.

In the first three years following diagnosis we were in and out of the hospital monthly (usually for 1 week at a time), because his central line would cause infections in his blood and those infections were only treated by IV antibiotics that had to be administered in a hospital setting. We would follow him around with his IV pole so he could attempt to learn to walk again. We had him in early childhood intervention for PT, OT and Speech. We were doing everything we could to not only keep his body stable

but to make sure he would not fall behind mentally and physically.

The nurses that came into our home, the hospital staff that we got to know very well during our numerous visits, our families, and our friends were unbelievable. As they say, "it takes a village to raise a child" and they proved that to be the case. We couldn't have done it without them.

When Ethan was 3 years old, he decided one day that he wanted to try some of the food that we were eating. It was one of the best days of our lives. He hadn't eaten anything orally in such a long time and he was showing interest. We were so excited. We went and got everything we could think of that would taste "good" and set it all in front of him and he took turns licking each item. This was it. He had interest. We had hope. We now understood that it would get better. We were ready to take the next step to get him off of all of these tubes and move forward.

It was a long road. Between feeding team appointments, central line infections and hospital stays, lab work, doctor appointments, lack of sleep, working full time and so many other everyday things those years became a blur. It took us several years, but Ethan was eventually getting enough nourishment by mouth to stop using the feeding tube for nutrition and we only used it for meds. We were able to remove the central line because we were able to eventually stabilize his levels through liquid medications given through his g-tube and the vomiting had subsided. This was huge, a major milestone for our entire family, we no longer would need nursing care in our home, no longer had to have someone come take his labs three times a week, could go outside our home without having to have a bag attached to him



Ross, Jenni, Ethan, and Jackson Bartkowski in 2019.

everywhere we went, he could take a shower or bath without having to put layers of protection on his skin only to pray we would not get the line wet and cause yet another infection...he would be able to start to try and have a "normal" life.

From the age of five, Ethan has had a fairly normal life. Of course, he still needs copious amounts of medication multiple times a day, eye drops administered more often than any young child and parent would like, numerous doctor appointments and many other challenges related to cystinosis. However, Ethan has come a long way and has been medically stable for several years. In so many ways, he has thrived. He was recently told by his physical therapist that we could take a break from seeing her because he has come so far in the

last couple of months. Today he is often found playing and/or "fighting" with his younger brother Jackson (7), practicing his jump shot in the driveway, playing with his Grandma's dogs or finding every excuse as to why he should do his homework "later".

While there are far better ways to learn lessons in life, Ethan having cystinosis has taught our family so many priceless lessons about perseverance, compassion and acknowledging that things can always be worse. We are so grateful for our family and friends and all of the support they show us, day in and day out, and the wonderful people we have met on our journey with cystinosis. It hasn't always been easy and future challenges are likely to arise, but we will get through them together.

FAMILY STORIES: Dicks Family Story

By Jon and Shirley Dicks



CRN Family Conference.

It was a perfect, storybook pregnancy. My wife and I had discussed the possibility of having a home birth, utilizing midwives and having our first born in the loving familiarity of our own home. Everything was just so us... we did hypno-babies, learned how to keep and nurture our “positivity bubble” and taught us as a couple how to take an active role in the growth and subsequent birth. On May 20th, our “little guppy” came in grand fashion. Shirley progressed so fast and so smoothly that I almost assumed the role of midwife as our team arrived a mere 15 minutes before Elle was born.

Eleanor “Elle” was the perfect newborn, she rarely cried, and save for a tongue-tie she nursed well and was putting on weight, we were head over heels in love with this beautiful little girl. Around 10 months Elle came down with a stomach bug and it was after she got over this bug we noticed she wasn’t really swallowing any of the food she put in her mouth. At first, seeing her chew up food and

slowly spit it all out was funny and could have easily garnered a win on America’s Funniest Home Videos, but that got old quick. Elle nursed well but had a serious issue with swallowing. At first we were told it was “normal” and would just be a transient phase of limited growth, but that was the main problem, she wasn’t growing. At her 12-month appointment her physical was completely unremarkable, she was very healthy, but just small. We continued to discuss her inability to swallow much. She was eating very little (hummus and yogurt were her staples), spitting most other foods out, drinking lots of water, and nursing on demand. No one seemed too concerned, she was highly advanced for her young age and had an ever-growing lexicon that placed her around 2.5 year old in speaking and comprehension. She was wanting to walk, constantly holding our hands, but could never find the strength to do it on her own. Her 15-month appointment was a low point, she was not gaining again. We pleaded with God to give us some answers because we were literally watching this bright star of ours begin to dim, ever so slightly, but her light was fading and we were running out of options.

We got a referral to OT/SP/feeding therapy for help with possible oral sensory issues and thus began our first introduction to CCHMC (Cincinnati Children’s Hospital Medical Center). December came and we knew something was gravely wrong, we failed our feeding therapy trial. Elle actually lost weight, our options got even slimmer. Nutritionists were consulted and added to our ever-

growing list of sub-specialties who all had one thing in common, no one knew what the hell was going on.

At this time GI became everyone’s sole focus. They kept calling it “failure to thrive” which we hated. The diagnosis is anything but, what child doesn’t want to “thrive”? “Are you kidding me?” I snapped at the specialist. I was upset, we were upset because the onus of her health was being pushed back on us as parents, and by proxy, felt like somehow this was Elle’s fault. As an Emergency Department critical care RN in a Level 1 trauma center, and a soon-to-be FNP-BC I know all too well that a diagnosis such as this is code for “we literally have no idea what is happening.” It’s a trash diagnosis that tunnel-vision providers into a plan of care that fails to look at the causality and the real question “why is this little thing not eating?”

As such the focus began to approach lunacy. First we were to blend up ice cream in everything she drank, of course that didn’t work. They ignored the fact that she only drank water and breast milk. The feeding team and GI told us she needed to eat more and to limit her water intake because they thought she was getting full off all the water she was drinking. December crept forward, Elle was fading fast. We were referred to consult Endocrinology for bloodwork to rule out everything else.

Most of us reading this can relate. We received a frantic call from a resident in the ED at CCHMC telling us we had to come in immediately and we would expect an admission STAT. Her electrolytes were way out of sorts, an IV and NG tube were placed as Elle

was extremely dehydrated. Mind you, no one would or could answer the questions as to why in someone who drinks copious amounts of water could present so dehydrated?

Cue the worst 8 months of Elle's life, we call this the "vomiting period." No matter what they pushed through her NG, she threw it up. She was constantly waking up the hospital room pleading for water. Again we were told to limit her water intake, they thought her stomach was getting too full on it, causing her to throw up her feeds. Formulas changed, rates changed, doses changed and still she was getting worse. She looked awful and felt awful. At one point she was screaming for water and the doctor looked at us and said, "it's a behavioral thing now." My wife looked at him dead in the eye's and screamed, "what child at this age screams for water just because?" This was our 5th night and we were over their plan of care. If all we were doing inpatient was titrating an NG feed, we determined this was something completely manageable at home. We requested an immediate discharge. Our team had failed to

identify anything of real value, and if we stayed another day, there was a serious chance a resident was getting headbutted. We went home late the night before Christmas Eve.

It was eight torturing months of unsuccessful NG tube nourishment. I will never forget the look of terror Elle had in her eyes as she would literally dry heave her NG tube out of her nose, and I had to re-insert it, sometimes up to four and five times every day. Until the day I die, I will always carry guilt for dragging my feet on failing to transition her sooner from an NG to a G-tube. Elle continued to throw up multiple times a day, her food intake continued to decline, and she was very weak. Formulas and rates continued to be changed to see what she could tolerate. A scope called an EGD was performed that once again was unremarkable for any ulcers, obstructions, or signs of disease. We were told, "sometimes we just don't know why kids won't eat."

It was finally decided to put in a G-tube so we could try a blended diet with her. In 2017, Elle had her second procedure to have it placed. I remember how happy I was that

we finally could get this damn tube out of her nose. At first it seemed this was the answer, she stopped all vomiting and was gaining... but only for a month. Then the cyclic vomiting episodes started again, we were at a loss.

Her water intake continued to increase as her energy continued to decline. She would get winded playing on the slide or simply climbing the stairs in our home and began refusing to go outside if it was sunny or hot. Despite it all, Elle's spirit was never stronger, she remained happy with the fiercest of personalities.

In our constant struggle for answers Elle had a swallow study, and a gastric emptying scan performed to determine a cause for the vomiting and feeding intolerance. We consulted an ENT specialist and pediatric dentist who all keyed in on a possible allergic etiology for her issues, as such and after much debate we had her tonsils and adenoids removed. After all these exploratory procedures we discovered a severe gastric delay, but still no one was connecting the dots. It felt like everyone thought their specialty was running the show, so no continuum



Our two-year-old.



Elle turns 4!



A smile that could light up any room.



A few uncles showing their love and support. Left to right: Brian, John, Jason with Elle, Tyler, and Joe.

of care was really established that looked at her presentation in the entire picture from birth to where she is now. She went on a trail medicine to help with the delay, it did nothing.

We were hitting hurdles at every direction, no answers. Still we pushed for more answers, and by this time our GI and feeding team visits were approaching the “frigid zone.” The tension in the room was always strained, no one would look us in the eye, they knew we were pissed. They sent us back to genetics at our request. Without blood work the doctor looked Elle over up and down and determined it was not a genetic issue, but a nutrition issue as she was perfectly proportional. She needed more calories.

Back to GI and still with no signs of improvement, we decided to try a GJ tube so we could get her extra calories overnight. The overnight feeds proved unsuccessful. She continued to throw up, would toss and turn, dry-heaving all night which left her miserable and exhausted most days. By her 3rd birthday she finally hit 20 pounds, but it was a long-fought battle to get there, and still we had

zero definitive answers.

We never knew what the next day would bring and we both were exhausted and frustrated. Through it all, Elle continued to be happy, sharp as a nail, and forever positive. She enjoyed going places and doing things as long as it wasn't too hot, that she had her sunglasses and plenty of ice cold water. She slowly crept up in weight, ounce by ounce. By August 2018, at yet another GI follow up, she basically gained a few ounces and still hadn't grown in height. Our GI doctor had no clue what to do next and basically asked us how we wanted to proceed, we decided we needed a reset. We would find that plan of care with endocrinology as GI was off the table now after 2+ years with no diagnosis and failing to even progress Eleanor out of the 1st percentile in height and weight. He stated to us “I just feel like I am failing you” we agreed and parted ways knowing something would have to drastically change moving forward. With endocrinology in mind we looked for endorsements on the best candidate to deal with Elle's unique feeding issues in concert with her overall

presentation. We needed someone to finally hear us and look at the big picture of her health history.

Philippe F. Backeljauw, MD came highly recommended as one of the world's most renowned pediatric endocrinologists with specialization in growth disorders. He was out on medical leave until October, but we both felt waiting for him specifically to see Elle was paramount. We were throwing all our faith into this visit, we had nothing left and Elle was suffering in silence.

A small man in size, slim, wearing wire-rimmed glasses who spoke in metered affect with the most mellifluous French accent came in to greet us. He shook my hand, then my wife and turned his eye towards Elle and shook her tiny little hand. He arranged himself in his seat and brought up all the lab work ever done on Elle up to that day and swiveled the screen to me and said “I'm very concerned about Eleanor.”

Finally, someone realized something was seriously wrong. “I believe she has been in metabolic acidosis for two years” he said. As we reviewed the

lab work, it became clear that Elle's care was gravely mismanaged with too many sub specialists, each taking a myopic view of their practice of care. In doing so they missed for 2.5 years what one man saw in 5 minutes of looking at her history.

My heart was beating out of my chest, as he continued. Eleanor, right on cue had to use the restroom and Dr. Backeljauw was insistent on a urine sample. We had a laugh as the nurse asked if we needed a hat to place in the toilet for collection and Elle shot back "no thanks, I can pee in the cup." She had potty trained herself at a very early age with all the frequent trips to the toilet throughout the day.

My wife Shirley and Elle went to provide the sample. With my head spinning Dr. Backeljauw and I continued to discuss her case. I had this crazy feeling as we were discussing the signs and symptoms she was presenting with, that I needed to have one pressing question that had always bothered me asked. I said "this may just be me flexing the dad muscle too strong, but I feel like Elle has an unnatural aversion to the sun." I watched as the color dropped from his face. His eyes got big, and by this time the girls had returned. We discussed the past minutes of conversation and Shirley also endorsed this aversion to the sun as well as her constant need for water in the presence of adequate water intake. Phillippe took his glasses off and said in a most serious tone "you brought this up to me, I didn't suggest this."

He turned to his computer and began to type, then turned the computer and showed us a few differentials with one that caught my eye as every symptom described Elle completely. He kept talking, I felt so lightheaded and



Celebrating Elle's 3rd birthday and reaching 20 pounds!

underwater that I had to tell myself to pay attention. Maybe it was shock, maybe it was a catharsis from three years of uncertainty to now being on the cusp of a true diagnosis, or maybe it was knowing how sick she truly was. I quickly snapped back to reality when he told me how concerned he was not for her gastrointestinal issues, but rather her kidneys. That day we got Elle's blood work redone, urine checked and a tentative diagnosis of cystinosis, the real ride was now just getting underway.

We wanted this, but not with this diagnosis. The clinician in me knew that things undoubtedly would get better because now we could take an actionable and focused approach to getting her feeling better daily, we just had no idea that we would be starting with her in such an advanced stage of kidney disease, no one did. In hindsight we know that Phillippe saved her life that day, without question.

Silently, Shirley and I knew we would be getting a frantic call. Like clockwork the lab alerted the ED to Elle's bloodwork and we immediately were told to come to the ED as soon



Elle before diagnosis (left) and one year later (right).



That's our girl!

as possible. We packed our bags and headed in. For some reason everything was different this time. Most notably, we were not the only one's concerned anymore. Bloodwork was redrawn, and a plan of care for Elle began, the one change was that GI was nowhere to be seen and nephrology was now steering the ship. At 1 am the morning Elle had her first ultrasound of her kidneys. Everything was falling into place and for the first time, in the throes of this new and brutal diagnosis we truly felt heard.

There were times we felt like Elle was being treated as a case study with many eager residents and specialists trickling into the room to see this rare diagnosis in person. It was our job to make sure that stopped ASAP. I



Halloween 2019.

am certain I hurt some well-meaning clinicians feelings after calling them out for poor bedside manner and the understanding that while yes this diagnosis was rare, you better make damn sure I never again here someone comment how “exciting it is to see this outside of a textbook.” To make matters more difficult Shirley was 6 months pregnant with our son, we received news that this disease was genetically inherited, and our unborn child had a 25% chance of being born with the same condition. Our world was shattered and slowly we were re-building into our “new normal.”

After an insane three days of sleepless nights we were discharged home with a plan and the stark realization that Elle at three years of age was in stage three of chronic kidney disease. Instantly I recalled back to times when I would wash her in the tub and she would

instinctively try and suck the water of the washcloth, we were now part of a small community, a community we never wanted to be a part of, but one we desperately knew we needed. It seemed like no one could even approach an adequate understanding of what a typical day looked like for Elle. No one realized what intractable nausea and vomiting really entailed. Elle still was not swallowing but her energy and overall disposition was like night and day once she went on the systemic electrolyte replacement therapy. We were for the first time making her feel better.

It wasn't until our first our patient visit with nephrology that we truly knew we were being well taken care of. As fate would have it our lead clinician in charge of her care was Stephanie Benoit, a quirky obsessive nephrologist who immediately fell in love with Elle. It just so happened that for the past four years I had been working hand in hand with her husband Justin in an emergency department literally next door. Small world? Fate had placed this angel in our plan of care and we immediately knew we were home when she looked at us both and said “it’s my ultimate goal to be the best cystinosis physician in the world.” She was all in, we were too. She encouraged us to seek out second opinions and has been working directly with Dr. Paul Grimm who saw Elle in clinic all the way out in Palo Alto at his world renowned cystinosis clinic.

From that day forward Elle began to blossom and for the first time we were seeing the true version of our child. A child no longer in metabolic acidosis, who’s kidney disease was being well managed. We were just getting started and already Elle was progressing better than she had ever in her short three years of being alive.

The road forward has been anything but typical and linear, nor has it been easy for Elle. At first it meant a metered and repeated battery of tests to get her baseline lab work corrected as the mismanagement of her care caused lasting and chronic kidney disease that will require Elle to get a transplant at a very early age. Countless hours of debate between our insurance provider and concierge pharmacies needing prior authorization, with constant denials until an appropriate diagnosis could be crafted that allowed her to receive the many medications that were and are literally keeping her alive. As such she is now on a total of eight different base medications.

We see a speech pathologist for feeding and occupational therapies, a pediatric nephrology nutritionist as Elle still has a severe swallowing aversion with a hyperactive gag reflex, physical therapy to keep her strong and to continually manage concern for rickets and long bone abnormalities. She is on a 14-hour daily GJ feed with a smoothie we get delivered and then add all the medicine directly to. CRT (cysteine reducing therapy) consists of about 12 daily cystagon caps, with daily cysteamine eye drops to make sure she is clearing the corneal crystals. There have been some setbacks as her cyclic vomiting meant we had to come back into the hospital for a five-day feeding therapy trial to get her set for her home GJ feeding schedule.

I was blessed to attend my first Cystinosis Town Hall event this past year in Arizona that was funded by Horizon Therapeutics. Since Shirley was so far pregnant and at that time Elle was still in the process of normalizing her levels, I had to travel alone. I knew I was home when I timidly walked into the conference

room the first day and three little guys with their shirts off came blazing around the corner, one little boy screamed out loud “look dad they have a button too” as he ripped his shirt off and proudly showed off his hardware for his new friends to see.

After our son Findley was born we heard about the CRN’s conference in Philadelphia. We packed up the family and made the long drive. We have been welcomed into this incredible community of like-minded caregivers, patients, and leading clinicians from all over the world. Elle was the life of the party in Philly. I was simply trying to get a handle on all the love I felt from “strangers who instantly felt like family.” Seeing all those thriving with this disease was almost too much to bear as we finally met individuals and families living this same story before us who made it through to a good place. We are slowly getting to that place, and it is in times such as this that we reflect back on how far Elle has come in the face of seemingly insurmountable odds.

Elle is now approaching 27 pounds, meaning since she was diagnosed and began her CRT with the correct cocktail of electrolytes and adequate nutrition schedule has put on six pounds in just under six months. Her hair has begun growing again and she is living such a wonderful life despite all she deals with on a daily basis. We still are patiently waiting for her to ditch the GJ tube, but that takes time and patience. For now, we are on the “gain train” with a focus on her nutrition and swallowing aversion. If you’re wondering about Finn, he was screened at 3 weeks old and we were informed he showed no signs of active disease process, nor was he a carrier. He is growing like a weed, adores his big sister, and the two of them are inseparable. The two are working hard



Little but fierce.

to be on opposite sleep schedules, and while Finn is not talking yet, he also manages to always crawl in the opposite direction of Elle. He does however influence Elle and her want to swallow more foods in the most incredible of ways.

We continue to take each day dose by dose. She anxiously waits for her pump to beep signifying it is finished and she can get in a hot steamy shower. Her personality has grown in so many ways; she loves to sing, dance, and tell jokes. She is 4 months strong into ballet/dance and is loving every minute of it. She continues to prove to us every day

just how strong and brave she is. At her last GJ tube change there were no tears, just questions to the doctor about everything he was doing. She no longer puts up a fight to get her growth hormone shot. She sits on mommy’s lap and shows her where to give the prick and lets out a quick “it hurts.” It has been a year since the diagnosis, and we have now graduated to three-month nephrology appointments with even fewer blood draws in between. While the future holds much uncertainty there is one thing we can bank on, Eleanor may be little, but she is undoubtedly fierce.

CRN Awards Two Academic Scholarships

CRN wishes to congratulate the 2019 Scholarship winners. **Gracie Smith** was awarded the Individual with Cystinosis Scholarship. She is attending Abraham Baldwin Agricultural College in Tifton, Georgia, where she is studying nursing. She hopes to become a pediatric nurse. Growing up with the challenges of cystinosis has provided with empathy to meet the needs of ill children physically and socially.

The recipient of the Sierra Woodward Sibling Scholarship Award was **Taylee Julian**. She is attending Southern Illinois University in Edwardsville, where she is studying dental medicine to embark upon a career as a dentist. Growing up as a sibling of an individual with cystinosis she learned early she wanted to help people and pursue a career in the medical field.



Gracie Smith.



Taylee Julian.

Deanna Lynn Potts Scholarship Awarded

The Deanna Lynn Potts Scholarship was awarded to **Christina Morris**. She is attending Charleston Southern University in Charleston, South Carolina. Christina is pursuing a career in nursing. Her decision to choose nursing came from the caring nurses and doctors she encountered during frequent hospitalizations throughout her life. She hopes to eventually to continue her education in anesthesiology or become a physician's assistant.

The Deanna Lynn Potts Scholarship will now be offered through the Cystinosis Research Network. This transition is taking place to better serve our young adults with cystinosis. Information about this scholarship is available on the [CRN website](#).

Our sincere best wishes to these amazing young ladies.



Christina Morris.

Honoring #GreatGivers on Giving Tuesday

The CRN has been a proud participant in Giving Tuesday since 2015. Giving Tuesday is a global movement encouraging people to “do good.” The event has inspired millions to give, collaborate, and celebrate generosity.

This year, we asked the community to reflect on those who’ve helped along the cystinosis journey. Leading up to Giving Tuesday (December 3) we shared these experiences to recognized the sacrifice, generosity, and selflessness many of us have been fortunate to benefit from. A nod to those #GreatGivers.



Gail Potts honors daughter Deanna Lynn.



Jenn Loglisci (right) with Dr. Gahl (left).



Jana Healy (right) with sister Jennifer.

Our efforts surpassed the \$5,000 fundraising goal! A special thanks to each person who opened up and shared their #GreatGiver story along with our donors and contributors. We are still accepting volunteers. If you'd like to learn more [visit us](#).

Full Great Givers stories available [here](#).



Cheryl Simoens (left) with her father.



Victor Gardener and his Pastor.



Sara Healy (right) and Amanda (left).



Steve Schleuder and friends.

First Annual Miles for Moose Fundraiser

By Andrea Carr

About a year and a half ago I was struggling with watching my beautiful grandson suffer with this terrible, incurable disease. It was so hard to know that there was NOTHING that could be done other than treat this symptoms and do our best to keep him going day to day. His mommy, Larissa, tirelessly is his BIGGEST ally. She knows his schedule, his meds, his appointments, etc. like the back of her hand. She is amazing. These kiddos are warriors...but my goodness, their parents are just as incredible! So, as a grandparent, I was just feeling helpless. I did what I could to help mommy and daddy. I bought lots of diapers and wipes because Moose (Benson) would pee so often that they were always running low. I tried to find clothes that would fit him best and work best with the G-tube in his belly so they'd have quick and easy access to it. I would take big sister Lilly places because I know she needed a little extra attention sometimes. And one day, getting close to Cystinosis Awareness Day, I saw a post from this man, Clinton Moore, who was training for his 57 mile walk. It caught my attention. I had thought about having a fundraiser months before when Moose was first diagnosed but I wasn't really sure how effective that would be. I myself, am a former race athlete, so this was particularly interesting to me because I miss races. But Clinton, he was walking it himself! IN ONE DAY! Well...he's crazy. But in such an inspiring and incredible way. Not only is he all of those things, but he's the father of a cystinosis warrior himself, and it turns out...the President of the Cystinosis Research Network. This,

mind you, I didn't know until MUCH later.

This clicked in my head. I followed Clinton on the day of his walk. I cheered him on from my seat and I cried when he finished. And I just knew. This is what we need to do, but we need to involve EVERYONE. I know I physically can't walk that far and I know a lot of people can't. But maybe if we did it as a team, a RELAY. And so that is what it became. I reached out to Moose's mom and told her my idea. We then included her mom, sisters, and best friend and our committee was formed. I started the [Miles for Moose Facebook page](#) and reached out to the CRN to see how I could gain their support. When Clinton Moore reached out to me personally, I was almost star struck. I am honored to have CRN, Clinton and the support of our community walk with us on May 9th and hopefully every year after that until a cure is found. Moose is the most special little boy, and we would all be lost without him. And I just know that every other cystinosis family feels exactly the same way. Every time we hear of another loss in the cystinosis community, our hearts just ache for those families. They ache for us too. We may not all know each other personally, but we are all connected.

Our goal is to raise as many funds as we can with an initial target of \$5,700. The walk will be a full day relay event with teams of up to 14 people walking/running a total of 57 miles. It's a fun day, so no pressure. I want it to be very family oriented, music, food, etc. Children's Miracle Network will also be there, they have helped our Benson "Moose" and his family so



Benson aka "Moose" hooked up to his formula bag.



Moose manning the controls.

much...and they jumped at the chance to be a part of the walk. At this point, we just want to form as many teams as I can and get as many sponsors as we can to make this an incredible, annual event.

[Click here](#) for our donation page.

Email Andrea for details at milesformoose57@gmail.com.

Research Update

By Christy Greeley, Vice President of Research



Christy and Jack Greeley.

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 – Interim 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

Krishnan Padmanabhan, PhD
Assistant Professor
Neural Circuits and Computation
Laboratory
University of Rochester School of
Medicine and Dentistry

The Cystinosis Research Network's generous support has allowed for two important studies that will inform future work using the Ctns -/- knockout mouse. Both projects and the resultant publications are under peer-review, and we have acknowledged the support of the Cystinosis Research Network in both.

First, mouse models have been used to study the biological underpinnings of a number of neurological and neurodevelopmental disorders. Interestingly, most studies have exclusively used male mice, and therefore there are no baseline measures about differences between males and females for a number of experimental procedures. As cystinosis affects both sexes, it is essential that experimental studies using animal models reflect this. In this work, we found that there are differences in behavioral habituation between control male and female mice on a run wheel, which is often used to study cognitive function and decision making. Specifically, we found that female mice run further and faster early during the habituation period, revealing sex specific differences that could inform our understanding of hormonal circuits, neuromodulators, and the systems involved in sensory processing.

In parallel, the lab is interested in understanding how neural circuit function is altered in animal models where a specific cellular phenotype has been implicated. The challenge is that neural activity originating from the brain is highly complex. For example, a few hundred cells (a small subset of 100 billion neurons that make up the human brain) have the capacity to represent over 10^{100} different patterns of activity; this number is larger than the number of atoms in the universe. As a result, new computational methods are needed to measure the activity of the brain and to understand how that activity reflects sensory coding, behavior, and motor action.

Without accurate tools for measurement and analysis, progress cannot be made in understanding disease processes. In a second project, we have imported methods for the area of statistical physics, using a mathematical framework called maximum entropy modeling to assess changes in brain activity. Specifically, we identified how the global activity of neural networks changes with behavior including how the interactions between neurons constrain the possible patterns of activity. We also uncovered how certain behaviors, such as running, change these interactions, and thus influence the structure of neural activity. Finally, we identified how changes in the circuit lead to alterations in the interactions between neurons, and thus the global structure of that activity. Our approach in this domain will provide new tools and measures for assessing neural activity in both animal models of cystinosis, but going further, may provide ways to link mechanistic changes in brain structures with changes in activity patterns and behavior.



Christy, Grandma Arndt, Jack and Alex Greeley.

Mechanisms Underlying Neurocognitive Changes in Cystinosis – Interim Report

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

At this time, we have a short (and hopeful!) update on the developmental path of basic auditory processing and sensory memory in cystinosis. While cystinosis' impact on brain function is relatively mild compared to its effects on other organs, the increased lifespan of this population and thus potential for productive societal contributions have led to increased interest on the effects on brain function. Nevertheless, and despite some evidence of structural differences, the neural impact of the mutation is still not well characterized.

We tested basic auditory processing

in a group of 36 individuals with cystinosis (6-38 years old) and in neurotypical age-matched controls (n=39). High-density electrophysiology (EEG) was recorded while participants were presented with a passive duration oddball paradigm using three different presentation rates (representing different levels of demand on memory). We examined whether the N1 (basic auditory processing) and mismatch negativity (MMN; sensory memory) significantly differed between groups, and characterized the developmental trajectory of these processes in cystinosis. Individuals with cystinosis presented similar N1 responses to their age-matched peers, indicating typical basic auditory processing in this population. The MMN response, however, was clearly reduced in the longer presentation rates in the children and adolescents, whereas the adults presented similar responses to the neurotypical controls. These findings suggest shorter lasting auditory sensory memory traces, and thus a sensory memory impairment in younger patients, which seems to



Community Advisory Board members, Dublin November 2019.

be resolved by adulthood. More work addressing other aspects of sensory and working memory is needed to better understand the bases of the differences described here and their implications.

If you would like to know more about these and other findings, please contact me at ana.alvesfrancisco@einstein.yu.edu. Though this project has come to an end, there are still plenty of findings to be reported. We will continue to update you on those as the data, now that we have recruited enough adults with cystinosis, are analyzed. We would like to wholeheartedly thank all the individuals and families that participated in our study and the Cystinosis Research Network, for so generously funding this project. Working with each one of you was an immense pleasure and a constant source of inspiration!

COMMUNITY ADVISORY BOARD

I am honored to participate this year as the U.S. representative in the EuroCAB programme, a project of EURORDIS, the European Rare

Disorder Organization.

The Community Advisory Board's (CAB's) objective is to improve patient access to novel therapies and treatments. This is achieved by engaging with clinical trial sponsors at the earliest stages of their research processes. The CAB also works with pharmaceutical companies on topics like educational materials and other appropriate topics. As well as meeting with industry sponsors, the Board engages with early-stage researchers as part of PPI - Public and Patient Involvement in research.

The first meeting of the Worldwide Cystinosis CAB was held in Dublin, Ireland at the end of October and early November and included representatives from Ireland, Northern Ireland, Spain, France, the Netherlands and Belgium, Germany, the United Kingdom, and the United States. These patient advocates reviewed research proposals and met with future sponsors of clinical trials to discuss their plans and provide expert input. The Worldwide Cystinosis CAB plans to meet again in July

2020, in advance of the International Cystinosis Conference.

EDUCATIONAL RESOURCES

All of CRN's educational materials including brochures, guides and other publications have been updated and are available on the CRN website: cystinosis.org.

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

Please visit the Research page on the CRN website for updates on CRN funded studies as well as other research from around the world: cystinosis.org/research/. Also be sure to check out the many cystinosis related articles and publications available in our [Publications and Guides library](#).

The Cystinosis Research Network, Inc. Financial Review — Accrual Basis

By Jenni Sexstone, Treasurer

For the 9 months ending September 30, 2019:

Revenues

Total income for the nine months ending September 30, 2019, was \$311,000 compared to \$378,000 in 2018 due to increased funds for the 2019 Family Conference in Philadelphia.

Expenses

Total expenses for the period were \$510,000 compared to the same period in 2018 of \$262,000. Education and conference spending was \$355,000 compared to \$92,000 in the prior year due to the biennial family conference in July 2019. Year-to-date research grant expenditures were \$107,000 compared to the same period in 2018 of \$123,000. Total operating expenses of \$45,000 were slightly higher than the same period in 2018 of \$36,000 due to website and database updates.

CRN had net operating loss of (\$197,000) for the nine months ending September 30, 2019 largely driven by the expenses related to the family conference. 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 \$196,000. Net change in cash through the third quarter of 2019 was a decrease of (\$165,000).



The Sexstone Family.



*Individuals living with cystinosis,
2019 Family Conference.*



Jordan and Jenni Sexstone.

Cystinosis Research Network and Amazon Smile

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

How do you do it?

1. Visit smile.amazon.com
2. Select your account
3. Under "Settings" select "Change Your Charity"
4. Type and select "Cystinosis Research Network"

amazon smile
You shop. Amazon gives.



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**

To request a program near you, please call 602-953-2552.



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

APPROVED USES and IMPORTANT SAFETY INFORMATION for PROCYSBI

What is PROCYSBI?

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 on the following page, and the PROCYSBI Full Prescribing Information available at PROCYSBI.com.

IMPORTANT SAFETY INFORMATION for PROCYSBI (CONTINUED)

What is the most important safety information I should know about 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, such as 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. 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. **Tell your doctor right away if you get a skin rash.**
- **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.

What should I tell my doctor before taking PROCYSBI?

Tell your doctor if you have any other medical conditions, including 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.
- 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.

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?

- See “**What is the most important information I should know about PROCYSBI?**”

The most common side effects of PROCYSBI include: vomiting, nausea, stomach (abdominal) pain, breath odor, diarrhea, skin odor, tiredness, skin rash, headache, problems with body salts or electrolytes.

These are not all of the possible side effects of PROCYSBI. Call your doctor for medical information about side effects.

For additional important safety information for PROCYSBI, contact your doctor, or review the Instructions for Use in the Full Prescribing Information, available at PROCYSBI.com.

You are encouraged to report negative side effects of prescription drugs to the FDA.

Visit <http://www.fda.gov/medwatch> or call **1-800-FDA-1088**.



Global Genes Rare Patient Summit: San Diego

By Clinton Moore, President

For the 2nd time, I had the opportunity to attend the Global Genes rare patient summit I San Diego in September. This is what I would consider the “Cadillac” of all the rare disease meetings held throughout the year by a number of organizations. With around 1000 attendees this year, this meeting is clearly growing, and at a rapid rate.

It's always an exciting time when you get to reconnect with friends you've made over the years from all different rare disease states. From listening to their successes, failures, and challenges they have had over the past year to sharing your own with them... we may all come from very different rare disease states but we all have something in common. One thing I've learned from attending these types of meeting is there are no strangers and everyone is interested in listening and sharing. Were all family. A rare family.

When attending this meeting I try to split it into 2 tasks.....

1. Attend sessions that will benefit my community, like how to better lead an advocacy group, how to increase fundraising, or understanding the insurance processes that seem to frustrate us all...and 2.... Attend sessions that will benefit me personally, like how to care for yourself while care giving, how to better speak with healthcare

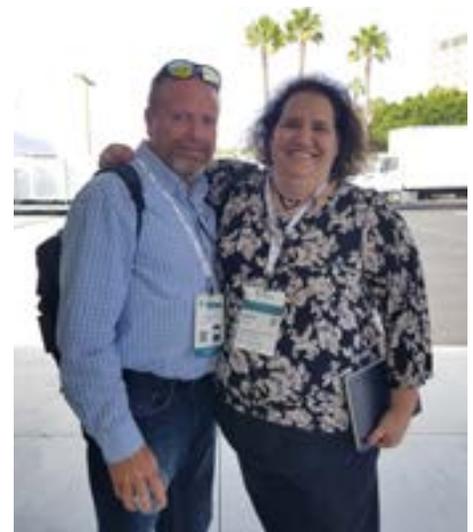


providers, or how to insure your getting the best possible healthcare for yourself or child.

Global Genes does an incredible job at covering a very broad spectrum of topics that affect us all in one way or another. It constantly amazes me how they can find ways to engage the audience and not just have speaker after speaker giving lectures in a language that is hard to absorb.

For me personally the time with other rare disease organizations, patients and caregivers is by far my favorite. We come from all walks of life from all over the country, so we don't get to see or speak to each other very often. So this time is very special.

Industry also attends this meeting, so this is another opportunity that I take advantage of. Discussing a variety of topics (and often complaining) face to face is very beneficial. The best way to work with industry to have any type



Clinton Moore and Carrie Ostrea.

of relationship and to attempt to keep them treating our community fairly is to face them head on.

All of the above things can be accomplished at this one meeting. Looking forward to attending again next year to continue learning, educating, and building relationships.

2019 RARE Patient Advocacy Summit: Global Genes Hosts Cystinosis Advocates

By Natasha Faisca

In September I attended my very first Global Genes conference in San Diego. I chose to attend Global Genes to learn how to be a better advocate for my children, and maybe help others in our small and growing cystinosis community.

The amount of people that attended alongside me was amazing! Over a thousand attendees. To learn that there are not hundreds, but thousands of rare diseases just blew me away. The sessions offered covered everything from family planning to how to navigate social security (which I

know for a fact I'm not the only one who is struggling with it).

At the end of the week, I walked away with a strong drive to dive into the rare disease community; to raise awareness not only for cystinosis but for all rare diseases.

I have so much more knowledge on how to navigate insurance companies, how to be a better advocate for my children and how to communicate better with doctors. I also have a new perspective on the rare disease world and I want to be a part of the fight that is going on globally.



Clinton Moore and Natasha Faisca.

Save the Date
September 21-23, 2020

**Global Genes Rare
Patient Advocacy Summit**
San Diego, California

We are saddened at the passing of some of our cystinosis warriors this year.

They live on in our hearts.

- ♥ Samantha Grover
- ♥ Mason Roland
- ♥ Karley Prince
- ♥ Tiffany Araújo
- ♥ Lyndsey Beeler
- ♥ Laura McGinnis
- ♥ Ana Caroline Silva Venâncio



Join the Cystinosis Research Network

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

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

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

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

Joining the Cystinosis Research Network enables you to:

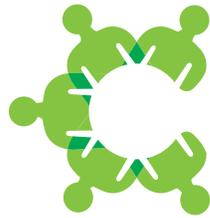
- Receive all the latest cystinosis information through our countless resources, including the biannual CRN Newsletter, our very informative website www.cystinosis.org, the popular online Cystinosis Facebook Support Groups, and our toll free number (1-866-276-3669).
- Attend the CRN Family Conference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

Join Cystinosis Research Network today!

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

Jen Wyman

VP Family Support, Cystinosis Research Network



CYSTINOSIS RESEARCH NETWORK

Join the Cystinosis Research Network Today!

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

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

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

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

Name:

Street:

City & State:

Zip Code:

Phone:

Fax:

Email:

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

Support CRN's Mission with Your Donation

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

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

Name: _____

Street: _____

City & State: _____

Zip Code: _____

Phone: _____

Fax: _____

Email: _____

In Honor of: _____

In Memory of: _____

You may send notification of my gift to: _____

Please check all that apply:

Friend

Parent of Child with Cystinosis

Family

Individual with Cystinosis

Professional

I am interested in volunteering for CRN.
Please contact me.

Search the Web with GoodSearch & Raise Money for CRN

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

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

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



Make Purchases at GoodShop & Raise Money for CRN

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

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

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





United Way Contribution Guidelines

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

Agency Name: **The Cystinosis Research Network, Inc.**

Non-Profit Tax ID: **04-3323789**

Address: **302 Whytegate Ct., Lake Forest, IL 60045**

Telephone: **1-847-735-0471**

Fax: **847-235-2773**

Email Address: **CRN@cystinosis.org**

Website: **www.cystinosis.org**

The local United Way organization will contact The Cystinosis Research Network via phone, fax, or e-mail to request we prepare and submit documentation verifying our status as a non-profit organization.

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

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

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

Donate to CRN by Selling on eBay

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



[Click here](#) to learn more about the CRN Amazon Smile Program.

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



CYSTINOSIS
RESEARCH NETWORK

Volume 12, Issue 2
Fall/Winter 2019

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Fax 847-235-2773

Email info@cystinosis.org

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.