CYSTINOSIS RESEARCH NETWORK

Volume 16, Issue 2
Winter/Spring 2024

THE CYSTINOSIS Advocate

INSIDE THIS ISSUE

SAVE THE DATE!

2025 CRN Family Conference

Amway Grand Hotel
Grand Rapids, Michigan
July 11 -13, 2025

Rare Disease Week 2024

#RareDC2024 Recap

700+ Rare Disease Advocates

207 Patient Organizations

332 Meetings with Members of Congress

Together we are many, together we are strong, together we are heard.
Rare disease advocates in Washington D.C. for Rare Disease Week 2024
(see more highlights on Page 4).

The President's Letter

Since our last communication in the Fall, the Cystinosis Research Network (CRN) has persistently pursued our vision for the future. Rooted in our mission are three guiding principles: advancing the search for a cure, enhancing treatments, and improving the lives of those affected by cystinosis. As we eagerly anticipate the 7th annual celebration of Cystinosis Awareness Day on May 7th, along with the planned in-person Family Meet-Up in Cincinnati, OH on May 4th, our enthusiasm knows no bounds.

Even in this off-conference year, the momentum within CRN is tangible as we embark on numerous initiatives. From pioneering innovative platforms, to leading valuable programs, we collaborate closely with esteemed industry partners to drive cystinosis research and support forward. Our unwavering commitment to funding new research studies is making a significant impact, leading us closer to curative therapies, now poised for pediatric trials. Despite the absence of our traditional conference, our dedication to fostering positive change in cystinosis care remains steadfast. Together, we move forward with renewed determination, empowering our community and shaping a future filled with hope.

We are excited to announce details of our groundbreaking medical symposium at The New York Academy of Medicine in May, organized by CRN. This event aims to address a crucial aspect of cystinosis care: the transition from pediatric to adult healthcare. Often, individuals navigating this transition lack necessary support, facing unfamiliar

The Dicks family, Finn, Jonathan, Ellie and Shirley.

medical practitioners and specialists. Through informative talks and interactive panels, we aim to bridge this gap, ensuring seamless continuity of care and unwavering support for patients, caregivers, and parents. As we navigate the complexities of cystinosis together, CRN remains committed to offering hope and assistance, even in the darkest times.

This conference is tailored for healthcare providers across various specialties, including those who may not directly work with cystinosis patients but can benefit from insights into managing this rare condition. By showcasing successes and innovative approaches in treating cystinosis, we aim to provide a model for transformation in other rare and chronic disease sectors. Through collaborative learning and cross-disciplinary exchange, attendees will discover how strategies employed in cystinosis care can enhance patient outcomes in other medical fields (see Pages 16-17 for details).

CRN's dedication to its community is evident in our wide and varying array of programs, initiatives, and support systems, made possible by our devoted volunteers and staff.

The President's Letter, continued

Many of these individuals navigate the challenges of caring for loved ones with cystinosis or managing the condition themselves. Together, we share the common goal of consigning cystinosis to history while paving personalized paths towards that future.

Each person touched by cystinosis brings a unique story and perspective, reflecting a diverse tapestry of backgrounds, languages, and journeys. Through CRN, families find solidarity in shared experiences and a supportive community offering guidance, camaraderie, and access to essential resources.

In the face of uncertainty, CRN stands as a beacon of hope, guiding families through the turbulent waters of cystinosis with unwavering support and a clear direction. As we journey together, we remain steadfast in our commitment to innovation, constantly seeking new ways to uplift our rare community, strengthen our bonds, and tirelessly advocate for our needs.

With deepest gratitude and honor,

Jonathan Dicks

Global Genes RARE Patient Advocacy Summit

By Megan Morrill, Director

I was honored to attend my very first Global Genes RARE Patient Advocacy Summit in San Diego this past September with Jonathan Dicks and Clair Johnstone. As an individual with Cystinosis, I was blown away by all the incredible organizations and like-minded individuals this event brought together that are dedicated to advocacy in the rare disease community. With over 600 attendees at the event, it was an honor to collaborate, connect, and learn from experts in the advocacy field.

Clair Johnstone, Jonathan Dicks, and Megan Morrill at the Global Genes Summit in San Diego.

The week started for Clair and I with a freshman orientation, where we were introduced to other first-time attendees. I was immediately impressed by the dedicated caregivers, individuals, and advocates working tirelessly to find treatments and cures for so many different rare diseases. We also attended a Global Advocacy Alliance meeting, a large networking event that allowed all to come together in one place for interactive programming for organizations like CRN to engage, learn, and build off one another.

The week continued with amazing presentations and discussions on many topics including coordinating complex care, developing scientific expertise to drive research strategy, developing sustainable funding models, and emerging rare policy issues. A highlight of my week were panel discussions, along with evening receptions where we had the opportunity to get to know all the amazing rare disease advocates on a more personal level. We learned about their passions, and what drew them to the rare disease community.

All in all, we left with a few more tips and connections to add in our rare toolkit, so CRN can continue advocating for Cystinosis Warriors. The week provided insight about the latest in rare disease innovations and advocacy, along with best practices for advocating on the individual and community level. For me, the week also provided continued hope and gratitude for how far the cystinosis community has come in regard to treatment, family support, and a hopeful cure on the way. Many in the rare disease community do not have these luxuries. Times spent at events like the Global Genes RARE Patient Advocacy Summit make me beyond grateful to be a part of such an amazing community.

Rare Disease Representatives from New Jersey meeting with Senator Booker's office.

Rare Disease Week on Capitol Hill 2024

Collaborative article by Jonathan Dicks, President, Clair Johstone, Executive Director, and Heather Rothrock, Director

The CRN proudly represented the cystinosis community at the 13th annual Rare Disease Week on Capitol Hill. Jonathan Dicks (President and VP Development), Clair Johnstone (Executive Director), Heather Rothrock (Board of Directors), and Gracie Smith (ALAB Representative) were in attendance along with rare disease advocates from all 50 states, Washington, D.C., and Puerto Rico. This year's theme – Unified. Amplified. Stronger than ever.

Day 1

Sunday began with a reception to connect with rare organizations like Global Genes, EveryLife Foundation (our event host) and smaller, disease-specific patient advocacy groups. The reception was followed by this year's rare disease showcase - a viewing of Bombardier Blood. This is the story of Chris Bombardier, a severe hemophiliac on his journey to summit Mount Everest. Fellow hemophiliac and filmmaker Patrick James Lynch chronicles the physical and emotional struggles of the summit, along with the added risk of Chris's rare disease, requiring frequent intravenous treatment to avoid bleeding episodes during the multi-day climb. While in Nepal, Chris's activism is spurred as he connects with other hemophiliacs, their families, and clinicians and learns of their disease experience in a developing nation without regular access to care and medication. The film also includes tidbits from Chris's other climbs as he worked toward achieving his pursuit of climbing the Seven Summits (the highest mountain on each continent) in an impressive six years. Though Cystinosis is physiologically different from Hemophilia, there are similarities that can be drawn between the exorbitant risk and complex medical needs of those with rare diseases attempting a journey like this.

> *"Having this medical condition didn't mean I was any less, it means that I'd already overcome a lot."*
> *- Chris Bombardier*

Day 2

The second conference day was a comprehensive working session, with presentations from rare disease advocacy groups and legislative professionals. Previous achievements of rare disease advocates were highlighted, such as amendments to the Orphan Drug Act and other highly relevant legislation and the proposal of a Rare Disease Center of Excellence within the FDA. With

Rare Disease Week on Capitol Hill 2024, continued

Patiently waiting for the official kick off of Rare Disease Week (Pictured left to right: Gracie Smith, Heather Rothrock, Jonathan Dicks).

the stage set for the impact advocacy can make, we delved into the details of this year's legislative priorities, or asks, that advocates would bring to legislators. For context around how these are selected, EveryLife commissions an advisory committee to review existing Congressional bills impacting the rare disease community and prioritizes those believed to be the best use of the advocates' time and resources in the current 118th Congress. Those selected for 2024 included asking Congressional leaders to:

1. Join the Rare Disease Congressional Caucus

Advocates urge state legislators to join the caucus, which aims to raise awareness among both the public and Congress about the unique needs of the rare disease community and its associated priorities.

2. Cosponsor the Accelerating Kids Access to Care Act
(Senate bill 2372; House bill 4758)

This bill focuses on expediting the process for providers to enroll in another state's Medicaid program, aimed at allowing rare disease Medicaid beneficiaries quicker access to specialists in other states.

Almost 50% of 2024 Rare Disease Week attendees said they've used out of state care.

3. Support the Safe Step Act
(Senate bill 652; House bill 2630)

Also referred to as "fail first." Currently, many patients with self-funded insurers and employer group health plans are required to try (and fail) medications preferred by the insurer before being approved for medications prescribed by their provider. This bill is part of the larger Pharmacy Benefit Manager reform package and aims specifically at requiring these plans to offer an expedient and medically reasonable step therapy exception process, enabling rare disease patients to start or continue treatment preferred and indicated by their provider without unnecessary interference from their insurance.

4. Renew the Pediatric Priority Review Voucher (PRV) program

Enacted in 2012, the PRV is meant to expedite development and promotion of drugs in specific communities, including pediatric rare disease, reducing the typical FDA approval process for these therapies from ten months to six. Given the program's impending expiration, Congress must pass an extension of the program prior to 9/30/24.

The majority of Priority Review Vouchers have been provided to rare pediatric diseases.

5. Support the establishment of an Interagency Coordinating Committee on Diseases

Establishing this committee would eliminate many of the silos that exist between HHS agencies like the FDA, NIH, NCATS, CMS, and others, allowing for a more streamlined, coordinated effort to ensure more effective and efficient resource utilization and stakeholder engagement.

Many patients, or 'lived experience experts' (as coined by aforementioned filmmaker Patrick James Lynch), along with other advocates offered compelling personal testaments related to each of the legislative asks and how the approval and/or establishment of these would positively impact the rare disease community as a whole. Later that day, attendees organized by state to meet advocates within their constituency and prepare for the next day's meetings with assigned House Representatives and Senators.

Rare Disease Week on Capitol Hill 2024, continued

"I was most excited about this breakout, and the chance to bond with other North Carolina natives on two things I'm most ardent towards - my love for my home state, and my passion for cystinosis. I have learned from our cystinosis community that complete strangers can share a bond almost immediately when it comes to rare disease- though the diseases themselves are all remarkably different, we all share an unwavering commonality of compassion for each other and an unspoken understanding of a life impacted by rare disease."
- Heather R.

Day 3

Following our working session, it was time for boots on the ground- day 3's meetings on Capitol Hill. The CRN crew had reviewed the legislative "asks" and were prepared to connect each one to its influence on the cystinosis community, as part of the greater conversation in each meeting with advocates and legislators. In total, CRN was represented in over a dozen different legislative meetings.

"The opportunity to advocate in this way so greatly represents the function of American democracy as intended, and on a topic that knows no political polarity. I was considerably impressed by the knowledge each congressional staff person had of the bills discussed and the thoughtful, meaningful dialogue in each meeting."
- Heather R.

With each state having two Senators, those meetings were typically larger groups with less opportunity to hear from each constituent. However, the House of Representative meetings were much smaller, allowing time to more specifically discuss the 'asks' from the Cystinosis perspective. Regardless of size, CRN left the legislative meetings feeling empowered and as though the Cystinosis voice had been heard and at a colossal, collective volume along with the other rare disease advocates.

All smiles from Heather Rothrock and the NC delegates speaking with Representative Foxx's office.

"If you are measuring impact by the number of attendees or passionate interactions, there is no doubt New Jersey made a substantial impression during our Congressional meetings. A standout moment was witnessing a 10-year-old share her rare disease odyssey; a task that can be intimidating at any age. It was a moment of pride, both as a NJ native and as an advocate for the larger rare disease community."
- Clair J.

Overall, CRN's time was well spent on Capitol Hill for Rare Disease Week in 2024. The group was able to get to know each other, telling stories and making memories while contributing to a greater cause through advocacy ultimately focused on improving life for those with Cystinosis and the rare disease collective. CRN will continue to follow these and other relevant legislative 'asks' with optimism that they will continue to gain momentum in Congress.

Visit the EveryLife Foundation for Rare Diseases' YouTube channel for a video summary of the event.

Rare Disease Day at the National Institutes of Health (NIH)

As the President, attending Rare Disease Day at NIH alongside my Executive Director was a deeply significant experience, especially following our intensive advocacy efforts on Capitol Hill over the past four days. Stepping into the event, we were met with a mix of emotions, ranging from exhaustion to excitement, but above all, a sense of purpose.

Throughout the day, we delved into discussions and presentations that highlighted the importance of this global observance. Rare Disease

Rare Disease Week on Capitol Hill 2024, continued

CRN President, Jonathan Dicks, with fellow Ohio advocates and Congressman Davidson's legislative aide.

Day serves as a stark reminder of the challenges faced by individuals living with rare diseases and the urgent need for collaborative action.

At the core of the event were discussions focused on the NIH's commitment to advancing rare diseases research. We engaged with the latest scientific breakthroughs and learned about NIH-supported initiatives aimed at developing diagnostics and treatments. These sessions provided valuable insights into the progress being made in our field and the potential impact on patients' lives.

Equally compelling were the opportunities to connect with fellow stakeholders within the rare diseases community. Engaging in dialogue allowed us to deepen our understanding of the complex issues at hand and identify areas for collective action.

The spotlight on patient stories was particularly moving. Listening to the voices of individuals living with rare diseases, their families, and their communities reinforced the significance of our advocacy efforts. These stories served as a poignant reminder of the real-life impact of our work and fueled our determination to drive positive change.

As we navigated through the event, we were struck by the diverse array of participants in attendance, representing various facets of the rare diseases ecosystem. From patients and caregivers to researchers and industry representatives, each person brought a unique perspective to the table, enriching the dialogue and fostering collaboration.

Rare Disease Day at NIH was not just an event; it was a testament to our collective commitment to advancing rare diseases research and improving the lives of those affected by these conditions. As we left the event, we carried with us a sense of continued purpose and a strengthened resolve to advance our advocacy efforts with unwavering dedication.

But wait, there's more! Please see the Adult Leadership Advisory Board section, Page 12 where you can hear from Gracie on her experiences during this year's Rare Disease Week.

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good days
effective compassion

Program Status:
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Assistance Amount:
$ 7,500 per year

Good Days is a national non-profit charitable organization that lifts the burdens of chronic illness through assistance, advocacy, and awareness.

We make life-saving and life-extending treatments affordable and act as your advocate, helping you navigate the system and guiding you to additional support through foundations and other organizations dedicated to those with specific, life-altering conditions.

Learn about our copay, travel and premium assistance programs to support individuals with Nephropathic Cystinosis.

Contact us for eligibility details. Ask a Good Days Patient Care Navigator for more information toll free at (855) 327-1134 or at www.mygooddays.org/apply.

Team USA at the opening ceremonies for the 12th Annual World Transplant Winter Games in Bormio, Italy.

WE DID IT! Right after completing the 5k cross country ski race with my dad Garry Hughes, waiting at the finish line with the American flag.

World Transplant Winter Games 2024

By Heidi Hughes

A few years ago, I came across the World Transplant Games through a fantastic group, the Chris Klug Foundation's webinar. At that moment, I excitedly declared to myself and my parents that I would compete in cross-country skiing at the next Winter Games. It has always been my lifelong goal to participate in an individual sporting event and prove to myself and the world that I am a worthy, strong, and determined person.

As I stood at the starting line (front row), surrounded by the stunning Italian Alps, fellow transplant recipients, and lifesaving donors, my dream was coming to fruition. Cinque, quattro, tre, due, uno… Go! This was it, everything I had prepared for, dreamed of, and strived for right at the tip of my skis. I pushed myself and my body to limits I never thought were possible.

Throughout the two events (5k and one-hour race) and multiple kilometers, my thoughts shifted to everything that I had survived to get to this point. I thought of the love and support of my parents, all of my Cystinosis friends, and the many we have lost, and my sheer desire to cross that finish line (twice) to show the world that I am a competitor.

> Every cell in my body was filled with strength, and I was determined to show that cystinosis WOULD NOT STOP ME from pursuing every single dream.

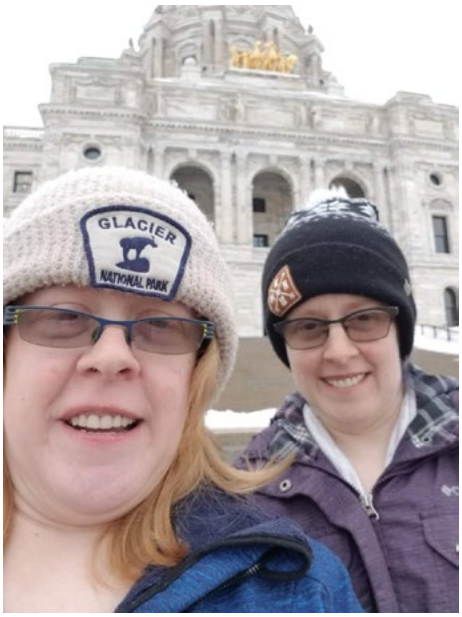
Instead, it has made me more resilient. These races were not easy by any means, and every competitor on that track was fighting for similar reasons. We all had the chance to show up and tell the world that the gift of life is what got us here, but our willpower and community of supporters backing us helped us cross the finish line.

This is my testament to the Cystinosis community; we can do anything we put our hearts and minds to. Never let the fear of failure or discouraging words from anyone stand in the way of your hopes and dreams. We are here because we continue to fight for our lives, and why not make that life unforgettable and powerful.

P.S. If anyone would like to get involved in events like this, please reach out. I will be participating in the future Summer and Winter games and would love to share events and accomplishments like this, with as many warriors as possible. This was a life-changing event, and I hope to help spark a fire for others in our community.

Navigating The Health Care World

By Sara Healy, ALAB Board Member & Jana Healy, Chair of ALAB



ALAB chair, Jana Healy with her sister Sara Healy, ALAB Board member continue to offer support the adult Cystinosis community.

We know the future with a rare disease can be uncertain. Everyone is in different stages of their rare disease journey. As young people, we think about our futures and the many choices and tradeoffs we need to make for our health and wellbeing. Choices such as selecting health insurance, understanding the limitations of disability coverage and social security support, along with the daily burdens and challenges of fighting for medication coverage and refilling medications, to name a few. Sara and I personally know how hard dealing with insurance can really be. Just getting our life-saving medications can be a chore. Every year my health insurance requires a prior authorization which consists of a letter from my doctor stating I need the medication to survive, otherwise it won't be covered. I should not have to revalidate that. Insurance companies should understand it is a lifetime drug.

This is one of the many examples of patients experiencing insurance challenges, and it is vital to obtain as much information as we can to best navigate this process. Our past meet up was titled, "Health Insurance Q and A". We had expert guests whose job it is to support patients navigating insurance challenges. They shared some valuable financial resources, disability support resources, as well as providing overviews on various types of insurance plans.

As you review these resources, if you have any questions with the information in our article, please email alab47213@gmail.com.

This information was provided by Samantha Sauer and Levi Peterson from the Patient Rising Helpline Insurance Information Sheet.

To purchase a health insurance plan, there are rules surrounding when you can and cannot purchase a plan. Open enrollment is usually in the fall through the beginning of the following year. Medicare Advantage enrollment dates usually fall within the first quarter of the year. The exceptions to these "rules" are: life changing events such as (marriage, divorce, or death in the family), sudden loss of insurance, change in address, had a baby (biological, adopt or foster), leaving incarceration, becoming a US citizen, etc.

Healthcare Plan Enrollment (https://www.healthcare.gov/get-coverage/) - use this link to apply through the ACA or help to find a local insurance navigator.

Enrollment Checklist (https://www.healthcare.gov/downloads/apply-for-or-renew-coverage.pdf) - helpful to get all your ducks in a row and make sure all your questions are answered.

Different Types of Plans:

HMO (Health Maintenance Organization): a type of health insurance plan that usually limits coverage to care from doctors who work for, or contract with the HMO. It generally won't cover out-of-network care except in an emergency. An HMO may require you to live or work within the service area to be eligible for coverage. HMOs often provide integrated care and focus on prevention and wellness.

PPO (Preferred Provider Organization): a type of health plan that contracts with medical providers,

such as hospitals and doctors, to create a network of participating providers. You pay less if you use providers that belong to the plan's network. You can use doctors, hospitals, and providers outside of the network for an additional cost.

POS (Point of Service): a type of plan in which you pay less if you use doctors, hospitals, hospitals and other healthcare providers that belong to the plan's network. POS plans also require you to get a referral from your primary care doctor in order to see a specialist. (HMO/PPO hybrid)

EPO (Exclusive Provider Organization): A managed care plan where services are covered only if you go to doctors, specialists or hospitals in the plan's network (except in an emergency).

Straight Medicare (Part A&B) - over 65 (there's a 7-month window after turning 65 to sign up for Medicare for the first time, or be qualified through disability (qualified for coverage two years after disability approval).

- Part A is hospital coverage only
- Part B is medical coverage only

Medicare Part C (Medicare Advantage Plan) - additional insurance coverage for anyone who qualifies for Medicare who can buy a plan of their choice through a Medicare approved private insurance.

Medicare Part D: prescription drug plan

Medigap: You must have Medicare A&B to purchase a Medigap plan. A Medigap policy only covers one person, so if you and your spouse both want Medigap coverage, you each have to buy your own policy.

If you have a Medigap policy and get care, Medicare will pay its share of the Medicare-approved amount for covered health care costs. Most Medigap insurance companies will get your Part B claim directly from Medicare. Your Medigap policy will then pay its share directly to your doctor and you're responsible for any cost left. Some plans also provide this for Part A as well.

If your Medigap insurance doesn't get your claims info directly from Medicare, you'll need to ask your doctors if they participate in Medicare; meaning they accept assignments for all Medicare patients. If the doctor participates, the Medigap company is required to pay the doctor directly if you ask them to.

Once you buy a policy, you'll keep it as long as you pay your Medigap premiums. All standardized Medigap policies are automatically renewed every year, even if you have health problems. Your Medigap insurance company can only drop you if:

- You stop paying your premiums
- You weren't truthful on the Medigap policy application
- The insurance company goes bankrupt or goes out of business

Medigap & Medicare Advantage Plans

A Medigap policy is different from a Medicare Advantage Plan (Part C). A Medicare Advantage Plan is another way to get your Medicare coverage besides Original Medicare. A Medigap policy is a supplement to Original Medicare coverage. When you're getting started with Medicare, you can either buy Medigap or enroll in a Medicare Advantage Plan, but you can't have both.

- If you have a Medicare Advantage Plan, you can't buy a Medigap policy. It's illegal for anyone to sell you a Medigap policy unless you're switching back to Original Medicare. If you want to switch to Original Medicare and buy a Medigap policy, contact your Medicare Advantage Plan to see if you're able to disenroll.
- If you have a Medigap policy and join a Medicare Advantage Plan for the first time, you may want to drop your Medigap policy because you'll be paying for coverage you can't use. If after you join a Medicare Advantage Plan for the first time and you're not happy with your plan, you'll have a single 12-month period (your trial right period) to get your Medigap policy back if the same insurance company still sells it once you return to Original Medicare. After that period, you might have to wait to drop your Medicare Advantage Plan, and you might not be able to buy a Medigap policy, or it may cost more.

Medicaid: Medicaid coverage is a state based program. Medicare and Medicaid can have similar qualifications, but guidelines and qualifications for Medicaid vary state-to-state. For detailed info, you'll need to go to your state's website.

What to consider when choosing a plan/changing current plan:

Medications. Have a list of what you take and what the coverage is for them. Different drugs are found on different tiers, and different tiers have

different co-pays.

Doctors. Make sure who you are already seeing, or planning to see is within the network. If you're on a Medicare Advantage plan, make sure the providers are still within the network. They may accept Medicare, but not Medicare Advantage. Same goes with non-Medicare insurance; make sure your providers are in network.

Costs. Medicare Part A is usually included and doesn't have a cost. That said, there are exceptions to that inclusion based on your SSI/ SSDI qualifications. Part B (for those who don't have any assistance from Medicaid or Medicare's extra help plan) is usually around $165. This changes yearly. Part C will have a monthly premium that you pay in addition to the Medicare Part B. That cost is determined by what private plan you choose and with what insurance company.

Make sure that you know the different costs that come with any insurance plan. **Coinsurance** is the percentage the insured has to pay after meeting a deductible. **Copay** is the set dollar amount you pay for visits, tests, surgery, hospital stays, etc. **Out of pocket maximum** is the most you pay out of pocket for the services per year. Once you reach it, insurance pays 100% of the services for the rest of the year.

Let's say your health insurance plan's allowed amount for an office visit is $100 and your coinsurance is 20%.

If you've paid your deductible: you pay 20% of $100, or $20. The insurance company pays the rest.

If you haven't met your deductible: You pay the full allowed amount: $100.

Pre-existing conditions: Medicare Advantage plans can be rejected due to having pre-existing conditions, or required to choose a Medigap plan for supplemental coverage instead. New York, Massachusetts, Maine and Connecticut are the only states that have protections against that.

Have a notebook and write down any and all information you can or have someone with you to take notes. If you have an insurance navigator, keep good notes of all conversations.

Other things to consider:

Dental/vision coverage. Unfortunately, neither of these are included in your "medical" coverage. They're under a different plan, or supplemental plan that might be included in your plan, but usually is not.

How much can you pay up front each month JUST to have coverage. High deductible plans, you pay less per month, but the deductible you need to hit per year is higher. Low deductible plans are more expensive per month, but there's a lower yearly deductible. These costs, on top of co-pay costs for visits, medications, etc., are all something to consider.

In addition, if you have pre-existing conditions, are the doctors you already see within the network for the plan you're choosing? What tiers do the medications you need fall under? What's the estimated cost per month for your health conditions within these plans?

ADULT LEADERSHIP ADVISORY BOARD

Apply Today!

Come help shape the future of our Adult Cystinosis support group. Together, we can build a better future for us all.

Apply online at https://cystinosis.org/alab

For more information, please contact us at alab47213@gmail.com

Explore More About ALAB

Check out our podcast: Cystinosis Rare: Journey into the Unknown

ALAB has monthly virtual meet-ups via Zoom - Check out our social media for dates and times!

Rare Disease Week on Capitol Hill 2024

By Gracie Smith, ALAB member

Gracie Smith with Georgia's Barry D. Laudermilk's staff.

When I was invited to go to D.C. for Rare Disease Week by myself, I was hesitant. If you've met me, you know that I am not much of a talker unless I really know you. So, it's safe to say attending something like this was intimidating for me. Nevertheless, I said yes. As the countdown began for advocating on Capitol Hill, the more my excitement rose. I had no idea at the time, but the experiences I would have, culminated into a week I'd never forget.

Sunday evening was the first event where all the attendees of the Everylife Foundation Rare Disease Week convened to watch a phenomenal movie, "Bombardier Blood". It was about the first person with hemophilia to climb the seven summits. I'm not a crier, but I was wiping away a tear or two during this one.

Monday came, and it came EARLY. People told me that this would be an incredible experience, but until I was in the middle of it all, I didn't know how true that was. During the first half of the day, we reviewed everything we would need to know going into our congressional meetings on Tuesday. We discussed the 'asks' in detail, and what each one entailed. I also heard some very moving personal patient stories throughout the day, which inspired us all in our advocacy efforts.

In the afternoon we joined others from our respective states. There were nine from Georgia including myself. Hearing everyone's introductions and journeys with rare diseases really put into perspective how different our experiences are, yet how the strong commonality of raising our voices for change, brings us together.

Monday evening I made my way to the YARR (Young Adult Rare Representatives) meetup. Mingling from one table to another, I found commonality with many other young adults outside of having rare diseases. We talked, laughed, and exchanged contacts. I hadn't realized an hour had gone by, and then it was time for everyone to part ways. I made some friends that night that I hope will last for many Rare Disease Weeks to come.

Tuesday came and the meetings with legislators began. We had a group small enough for everyone to share two minutes of their story. It was validating being asked genuinely inquisitive questions after telling them, in depth, about a part of my life that I've only shared with my closest circle. It was scary in a way, but knowing that by telling my story, I am potentially helping improve the health and experiences of future generations of people born with rare diseases like cystinosis, and it is a great feeling.

There were some directional challenges that our group had going from one white building to the next, navigating the streets of Washington D.C.! Nonetheless this added a little extra excitement to the day. After my last meeting was over, I took a deep breath. I DID IT!

This week was a whirlwind in the best way possible. I am so grateful to have gotten the opportunity to go to my first Rare Disease Week and represent the Cystinosis community. I couldn't have asked to go with a better group of people to this event. Heather, my new "Cystinosis aunt", was the entertainment, Jonathon was always making sure we were all pumped and ready, and Clair always had a plan and answered my endless questions about the event.

As I am sitting back at home after a week full of community, learning, and a lot of walking, I could not be filled with more gratitude. The Cystinosis community is small, and this week showed me that the rare disease community is also small, but big things often come from a small, but mighty group.

FINANCIAL RESOURCES

For Hospital Debt

https://forms.dollarfor.org/?aid=patientsrising
https://www.goodbill.com

For General/Miscellaneous Medical Expenses

https://accessiahealth.org
https://www.mygooddays.org
https://www.healthwellfoundation.org
https://www.resolvemedicalbills.com

For Co-pay and Medication Assistance

https://namapa.org/request-assistance
https://www.needymeds.org
https://www.patientadvocate.org
https://www.dispensaryofhope.org
https://www.rxhope.com
https://medicineassistancetool.org
https://www.goodpill.org
https://sirum.org/resources-for-individuals/
https://www.krogerspecialtypharmacy.com

For Disease Specific Grants

https://rarediseases.org/patient-assistance-programs/
https://tafcares.org
https://www.panfoundation.org
https://fundfinder.panfoundation.org

Patient Advocate Foundation (PAF)

Offers free case management to those dealing with chronic illnesses
Offer financial support via grants & co-pay assistance
Home - Patient Advocate Foundation
Call: 800-532-5274

Disability website: https://www.ssa.gov/disability
https://www.ssa.gov/disability/eligibility

Let's Keep in Touch!

ALAB will be having meet-ups once a month.

Stay tuned on social media for dates and times!

Search "Adult Leadership Advisory Board"

@cystinosisalab1

@cystinosisTEENS

Patient Services (RARE Concierge)

Our Patient Services team provides support to individuals, care partners, families, friends, and others impacted by rare diseases. We provide information, resources and connections based on your individual needs and living experience. Together with our partners, Global Genes helps people find and build communities, gain access to information and resources, and provide hope and support for the more than 400 million people affected by rare disease around the globe.

Our Patient Services Guides are dedicated individuals whose mission is to improve the quality of life for all rare disease patients, including the undiagnosed. They understand the wide array of concerns and challenges the rare disease community experiences.

How can we help?

CONTACT US

- Help getting a diagnosis
- Locate genetic counseling services
- Help find a doctor, specialist or center of excellence
- Provide information on financial & disability resources
- Connect to mental health & social services
- Caregiving resources
- Connect with rare disease community
- Connect to patient advocacy organizations
- Identify clinical trials & research studies
- Guidance on advocacy & raising awareness

Contacting the Global Genes Patient Services is **FREE** and available to all those affected by rare diseases.

Contact us:

patientservices@globalgenes.org

globalgenes.org/rare-concierge

Education & Awareness Update

By Marybeth Krummenacker, Vice President of Education & Awareness

I recently read this quote: “If I have learned anything in my decades as a journalist it is, that ONE person really can change the world” (Kristin Dahlgren, who retired from NBC to pursue her passion with the Pink Eraser Project. This is a collaboration of doctors and patients researching to discover a breast cancer vaccination). Then I had ONE of our wonderful Board Members, (thank you Karen Gledhill) send me two packages of historical documents relating to cystinosis. Boy did I have fun looking back on where the cystinosis community has come from. It was all because of ONE person, Jean Hotz, a grandmother who saw a need to make life better for the families living with this rare disease called cystinosis. Later, I saw a trailer for a movie called One Life, an extraordinary story of ONE man who helped rescue hundreds of predominantly Jewish children from Czechoslovakia. It was a race against time before the borders were closed by the Nazis during World War II. All of these themes were the same, it only takes ONE person to change a life!

I often think of our community of cystinosis as ONE; how we have been presented with opportunities, and are able to be a presence at various events. Jonathan Dicks, Clair Johnstone, Heather Rothrock and Gracie Smith all attended Rare Disease Day in Washington D. C. in late February, and represented cystinosis on the Hill. Laura Krummenacker, Emily Mello, Herberth, Jessica and Martina Sigler, and myself, all participated in Rare Disease Day at Quinnipiac University on February 20th. Carol and Garry Hughes, Terri Schleuder, Gail Potts, and Clair Johnstone, exhibited at the American Society of Nephrology Meeting last November in Philadelphia. We look forward to the upcoming Pediatric Academic Society meeting in Toronto in May.

Marybeth Krummencker and her grandchildren.

As I said in Nashville last July, we have such a history, and we have been so fortunate to have had so many wonderful people come into our rare disease world to help make life better for all of us, but things don’t just happen, we make them happen by showing up! We have a history of recognizing opportunity and making sure that cystinosis is a ‘voice at the table’. CRN represents cystinosis, the community of ONE, on so many levels. Ninety percent of life is just showing up, and that we continually do.

We are in the process of sponsoring and participating in a ONE-day event in NYC called ‘Cystinosis Symposium: A Rare Disease Model for Comprehensive Care’ at The New York Academy of Medicine. Doctors have been invited who are presently treating pediatric and adult patients to hear of the latest research, and to also engage in discussions and learn about treating cystinosis as a model for any rare disease to follow, from diagnosis to adult care. The goal of this meeting is to bring in specialists from the pediatric to the adult world, to come and listen and to learn what it is like to treat a rare disease like cystinosis. Dr. Frederick Kaskel has been instrumental in securing The New York Academy of Medicine and building the agenda for a full day of listening and learning from each other.

There are all kinds of opportunities within the Education and Awareness Committee. Find out what we are doing and continue to do to educate the world about our ONE disease, cystinosis.

CYSTINOSIS
RESEARCH NETWORK

CYSTINOSIS SYMPOSIUM

A Rare Disease Model for Comprehensive Care

SAVE THE DATE!

Friday, May 31, 2024 | The New York Academy of Medicine

1216 5th Ave, New York, NY 10029

Registration will be open to healthcare professionals with a desire to learn more about comprehensive care in rare disease. This one-day, in-person symposium will be held Friday, May 31, 2024 at The New York Academy of Medicine.

Review the latest advances in our understanding of cellular and molecular abnormalities in nephropathic cystinosis, which have led to three major breakthroughs. We'll explore the implementation of effective treatments for interrupting disease progression from childhood to adulthood, and the exciting potential of cutting-edge gene therapies to eradicate this lifelong condition. Despite these promising developments, substantial gaps and challenges persist in providing comprehensive care to cystinosis patients and families. Learn how earlier genetic recognition and timely diagnosis can make a difference. Find out how we're bridging the gap to construct multidisciplinary healthcare teams extending from primary care to specialized support, including social work, nutritionists, educators, and quality of life navigators.

Be a part of this transformative conference, one of the first to address these glaring deficiencies in cystinosis care. Together, we'll focus on themes that apply to healthcare providers, educators, and families, and work towards disseminating new information and raising awareness within the cystinosis community. Our ultimate goal is to provide an algorithm for more effectively navigating the promising future towards a cure. Don't miss out on this pivotal event!

REGISTER TODAY!

cystinosis.org/events **info@cystinosis.org**

CYSTINOSIS RESEARCH NETWORK

CYSTINOSIS SYMPOSIUM

A Rare Disease Model for Comprehensive Care

Friday, May 31, 2024 | The New York Academy of Medicine

1216 5th Ave, New York, NY 10029

All sessions located on the 3rd floor Library Reading Room unless otherwise noted. Agenda topics, speakers and timing is subject to change.

7:45a - 8:45a	**Registration Check-in and Breakfast**
8:35a -8:45a	**Welcome** – Frederick Kaskel MD, PhD, FAAP, FASN
8:45a - 9:15a	**Cystinosis: Basic Defect and Genetics** – Bill Gahl, MD, PhD
9:15a -9:45a	**Cell Biology of the mTOR Pathway in Cystinosis** – Jess Thoene, MD
9:45a - 10:15a	**Neuromarkers of Sensory and Cognitive Function in Cystinosis** – Sophie Molholm, PhD
10:15a-10:45a	**Gastrointestinal Issues in Cystinosis** – Adam C. Stein, MD
10:45a - 11:00a	**Break**
11:00a - 12:00p	**Panel: Patient and Caregivers, Challenges within Rare Disease** Jonathan Dicks, CRN President and VP Development, cystinosis caregiver Shannon Henderson, RN, living with cystinosis Christina Morris, Adult Leadership Advisory Board Member, living with cystinosis Herberth Sigler, CRN Board Member, cystinosis caregiver Joshua Zaritsky, MD, PhD (moderator)
12:00p - 12:15p	**Orthopedic Issues from Childhood to Adulthood** – Melinda S. Sharkey, MD
12:15p - 12:30p	**Clinical Trial Readiness for Myopathy and Dysphagia in Adults with Cystinosis** – Reza Seyedsadjadi,MD
12:30p - 1:00p	**Ophthalmic Concerns Related to Cystinosis** – Rachel Bishop, MD, MPH
1:00p - 2:00p	**Lunch** – Pharmaceutical Considerations and Collaboration in Rare Disease Leadiant Biosciences | Recordati Rare Diseases | Amgen Rare Disease
2:00p - 3:00p	**Panel: Transitioning from Pediatric to Adult Nephrology** James Drakakis, DO Ladan Golestaneh, MD, MS Larry Greenbaum, MD, PhD, FAAP (moderator) Rupesh Raina, MD, FACP, FAAP Cybele Ghossein, MD Jack Greeley, living with cystinosis Laura Krummenacker, living with cystinosis
3:00p - 3:30p	**Engagement and Support in Rare Disease: Cystinosis Research Network as a Model** Jonathan Dicks, CRN President, VP Development and cystinosis caregiver Marybeth Krummenacker, CRN VP Education & Awareness, cystinosis caregiver Kristina Sevel, MSN, RN, CRN VP Research, cystinosis caregiver Frederick Kaskel MD, PhD, FAAP, FASN (moderator)
3:30p - 3:45p	**Break**
3:45p - 4:15p	**Supporting Mental Health of People Living with Rare and Chronic Disease** Maya Doyle, MSW, PhD, LCSW Chelsea Meschke, LMSW, cystinosis caregiver
4:15p - 4:45p	**The Potential of Moderna mRNA Technology to Treat Cystinosis** – Paul Goodyer, MD
4:45p - 5:15p	**Novel Genetic Treatments of Rare Disease and Applications to Common Disease** John M. Greally, MD, PhD Melissa Wasserstein, MD
5:15p - 5:45p	**Stem Cell Gene Therapy Clinical Trial Results in Cystinosis** – Stephanie Cherqui, PhD
5:45p - 7:15p	**Wrap-up Reception** – Hors d'oeuvres and cocktails/beverages served *Located in the Periodicals Room, 3rd floor*

cystinosis.org/events **info@cystinosis.org**

A special thank you to our sponsors

AMGEN Rare Disease

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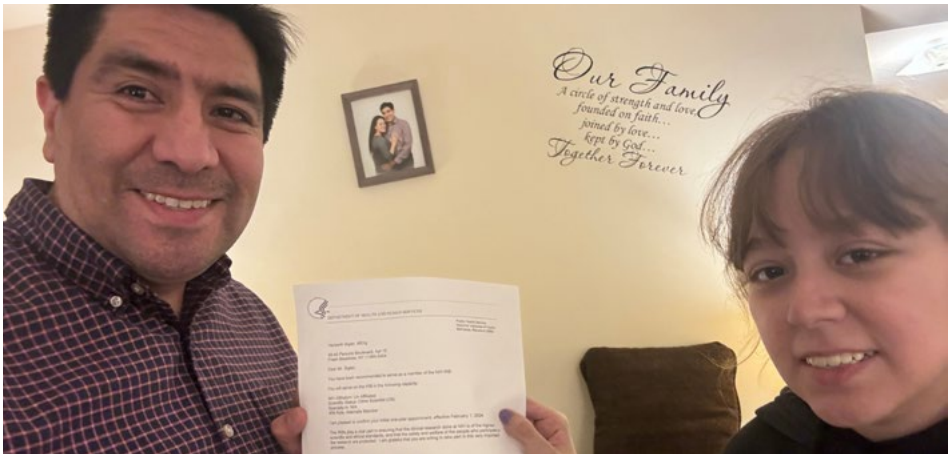

Herberth and Martina Sigler, celebrating his NIH IRB appointment.

Martina Sigler.

Herberth Sigler to Serve on the International Review Board at the NIH

By Herberth Sigler, Director

Hello CRN family, I am happy to share that I feel immensely honored and thankful for the opportunity to serve as a non-affiliated board member at the Institutional Review Board (IRB) at the National Institutes of Health (NIH). The appointment is for a year, and my time and dedication are ad honorem. This volunteer and pro bono position allows me to give back to our society and be part of the search for safer and better treatments for humanity. These new healthcare guidance and discoveries result from the complex and lengthy work of talented scientists and doctors who dedicate their lives to finding a cure or better treatments to improve our daily lives. My contribution here is being part of a group of people in our society who guard and protect a high standard of ethics and regulatory rules while human-subject research is happening.

An IRB is a result of a mandatory regulation in the US since the National Research Act was passed in 1974. The intent and goal of this law is to prevent abuses while research is conducted on human subjects. The creation of IRB boards applies to any US institution that does human-subject research. The IRB is responsible for providing ethical and regulatory oversight of human-subjects investigation. The board is divided into three types of members. The three parts must be represented and be part of the meeting to have a quorum. The scientific group is typically the primary investigator. The second group is the non-scientific group. They are not necessarily medical or health-specialized people, but those whose primary concern is other than science. Finally, a non-affiliated member of the institution that represents the society. These people are diverse members with little or no scientific or medical training or experience, who focus on ethics, when human subject research needs the approval to start, continue, or modify from the initial research.

I was introduced to the opportunity by reference when the IRB at the NIH searched for non-affiliated, non-scientific members. After Googling, reading, asking, and deciding to show interest, I needed to present my curriculum vitae, a survey, background check, not have financial or conflicts of any kind in being part of the IRB, go through training and mentorship, and attend meetings as an observer. Six to seven months later, here I am. I represent the majority of the human population that is not medically trained, non-affiliated to the institution, and has no expertise as an investigator, but I am a father of a child with Cystinosis. This child's life depends on medications that were the result of clinical trials and research at some point in the past.

To the CRN family and anyone reading this article, I feel profoundly honored to be part of this community and serve at the IRB. I will serve to the best of my capacity and knowledge in this unique learning opportunity. I will provide insight into the review of human-subjects research in this first year of my appointment. I am eager and willing to grow in this role and meet the high standard. Thank you, and I am looking forward to keeping the community posted.

Panel at the Rare Health Equity forum sponsored by Global Genes 2023.

Bridging the Gap: Rare Diseases, Scientific Research, and Health Equity

by Jonathan Dicks, President & Vice President of Development

Introduction

As the President of the Cystinosis Research Network, I had the privilege of attending the Rare Health Equity Forum organized by Global Genes. This event served as an eye-opening experience, shedding light on the intersection of rare diseases and the critical need for equity in scientific research and healthcare. In this review, I will reflect on the insights and discussions from the forum, emphasizing the significance of addressing health disparities among individuals affected by rare diseases.

Rare Diseases: A Hidden Challenge

Rare diseases collectively affect millions worldwide, yet they often remain hidden in the shadows of mainstream healthcare. The forum illuminated the struggles faced by individuals and families affected by rare conditions, from delayed diagnoses to limited treatment options. These challenges underscored the urgent need for a more inclusive and equitable healthcare landscape.

The Impact of Racism

One of the most profound takeaways from the forum was the undeniable impact of racism on healthcare outcomes. It was evident that racial disparities persist, affecting the diagnosis, treatment, and overall quality of life for individuals with rare diseases. Discrimination, implicit bias, and unequal access to care were acknowledged as significant barriers to equitable healthcare.

Promoting Diversity in Research

The forum emphasized the importance of diversity in scientific research. Inclusivity in clinical trials and research studies is vital to ensure that treatments and therapies are effective for all populations. Speakers underscored the need to actively involve underrepresented communities in research initiatives, empowering them to be part of the solution.

Policy Initiatives and Advocacy

The Rare Health Equity Forum highlighted the critical role of advocacy in driving change. It became evident that we must work together to influence policy changes that prioritize rare disease research and health equity. Legislative efforts and policy reform can have a profound impact on healthcare accessibility and affordability.

Collaboration is Key

A recurring theme throughout the forum was the power of collaboration. Stakeholders, including patients, advocates, healthcare professionals, researchers, and policymakers, must unite to effect meaningful change. Partnerships among organizations like ours, focused on rare diseases, are essential in advancing our shared mission.

Conclusion

The Rare Health Equity Forum served as a poignant reminder of the challenges faced by individuals with rare diseases, especially those from marginalized communities. It emphasized the urgent need for systemic change, challenging us to confront racism in healthcare and to strive for equity in research and treatment. As President of the Cystinosis Research Network, I am committed to amplifying these important messages and working tirelessly to ensure that our rare community receives the care and attention they deserve. Together, we can bridge the gap between rare diseases, scientific research, and health equity.

Savannah's Story

By Kelli Rupert

Hello! We are the Rupert family, and I am Kelli. I am honored to have the opportunity to share Savannah's story, and boy, does this little girl have a story! We'll start by back-tracking to the point when my husband and I decided to grow our little family. I had a daughter from a previous relationship, and Justin took to the stepfather role with grace and ease. I was so excited for him to experience having a baby as Annabelle was almost 4 years old when we met.

Kelli, Evelyn, Annabelle, and Justin holding Savannah.

Our journey to conceive was anything but easy, and after a year we decided to seek help from a fertility specialist. Fast-forward the story to several more months of failed attempts, and everything being placed on hold due to Covid. We knew our next step was IVF. Due to the financial burden of this route, we knew we likely had one shot, and decided to increase our odds of success by having multiple embryos implanted. Once we knew we were pregnant, the world felt lighter than it had in a long time. Then at our first ultrasound we found out we were having twins! Honestly, though, we weren't shocked, and had prepared ourselves for that possibility.

It was a very difficult pregnancy for me and brought on a lot of challenges. At our 20-week anatomy scan we found out that twin A (Savannah's twin sister, Evelyn) had a heart defect along with other issues with the pregnancy. This was devastating news for us and brought on a lot of fear and uncertainty. Several weeks later, as the pregnancy became more and more difficult, Savannah was diagnosed with intrauterine growth restriction, which meant there was too much fluid in her amniotic sac and it was restricting her from growing. Again, more fear, so much fear.

At 29 weeks, I went into preterm labor, and the girls were born at just 2 lbs 8 oz and 2 lbs 9 oz. We were prepared for the possibility that Evelyn's condition could decline rapidly after birth due to her heart defect, but for some reason Savannah was the one whose condition was much more critical, and had the medical staff baffled. It was suspected that Savannah had esophageal atresia which meant that her esophagus ended in a pouch and did not connect to her stomach. She also had tracheoesophageal fistula which meant that there was a connection between her trachea and her esophagus causing secretions to leak into her trachea that caused her to aspirate.

On day 2 of life, Savannah was transported to the Children's Hospital in Madison, WI and underwent surgery to place the stoma in her stomach (for later g-tube placement), and to clamp the fistula. At this point, we had two very premature babies with health issues at two different hospitals. It took two weeks and a team that fought hard for us, but we ended up getting Evelyn transferred to the Children's Hospital as well, so she and Savannah could be in the same NICU. They even made it so the girls were in the same room! This was such a huge relief for us, but we knew the girls still had a very long road ahead of them. Not much could happen until they grew and so that was the focus for quite a while.

We learned of some other physical anomalies that Savannah was born with, and as a result, we were paid a visit by the genetics team. They indicated to us that due to the combination of anomalies Savannah was born with, there was likely a genetic component. They wanted to run an extensive genetic screening on her. Of course we said yes, and never in a million years expected our lives could change so much. Once the results were back, the genetic team came to talk with us again, and explained the results turned up something we weren't even looking for. Yup, you guessed it, Cystinosis. I honestly can't even recall how I felt after receiving the news. I know it felt very serious, but all we could digest in

Savannah's Story, continued

Savannah Rupert.

-the -midst of everything else going on was that it was really good we caught it early on, and she needed to start treatment right away. By 2 months of age, Savannah was on Cystagon. After Savannah's surgery to connect her esophagus to her stomach, she could begin learning to drink from a bottle. She was a phenomenal eater! The twins were released from the NICU after 4 months and we had no idea what we were in store for.

The first year after the girls were born feels like such a blur, between all the surgeries and procedures, and what felt like never-ending new diagnoses for Savannah, (unilateral hearing loss, scoliosis, tethered spinal cord, etc.). We didn't have the capacity to fully understand what her Cystinosis diagnosis meant. It wasn't until around her first birthday when she suddenly started refusing bottles that we started learning more about this horrible disease. The internet can be such a scary place, but truthfully cystinosis is terrifying. I don't think there's any way the internet could sugar-coat it for us, and I found myself crying all the time trying to wrap my head around how we were going to manage everything.

Savannah was only in daycare part-time, and we had family helping out to fill the gap and help with mid-day medications. At this point Savannah was no longer eating orally. We switched to solely g-tube feeds where we had previously only been using it for medication administration. This also meant we had to pull her from daycare, as our in-home provider was not comfortable with the g-tube. Thankfully we had family that were in a position to be able to assume her care full-time, but after a while this also began straining our relationship with family.

Trying to find care for a medically complex child is beyond challenging, especially in a community where finding a daycare opening for a healthy child is already nearly impossible. By some stroke of luck, we were able to find a family who was willing to do a nanny-share with us and the nanny even had previous experience using a g-tube! Savannah was finally in a place where she thrived. It was also during a time when we struggled the most with managing her ever-changing medications and feeding schedules. We tried to find that sweet spot, where she wasn't feeling ill, and could keep her medications and food in her stomach. We sought out therapies as Savannah was delayed in her gross-motor development, and also had a severe oral aversion that stemmed from medical trauma and all the vomiting. I know fellow cystinosis families relate all too well.

We struggle the most with getting Savannah to grow, and just recently had a consultation with endocrinology to start the conversation about growth hormones. We have so many questions and are so incredibly thankful to have found our Cystinosis family that we can lean on for answers and advice. Savannah is an absolute light in this world. She has such a fun-loving and goofy personality.

Happy Birthday Savannah and Evelyn.

Savannah's Story, continued

Everywhere she goes, people are enamored by her. Whether it's her nearly white-blonde hair, her infectious smile, the sunglasses she refuses to take off even when inside, or the way she says "get outta town". Every time we feel exhausted and want nothing more than to have a bit of reprieve, we are reminded that Savannah doesn't have that option. She continues to fight day in and day out without any understanding. We cannot give up because she never gives up.

We feel like we live in a state of fear and grief at all times. Grief for how we imagined our children would grow up, grief for all the things we wanted for our family, and intense fear for the unknown and what the future will look like for Savannah. We have more hope than ever with the stem-cell trial, but know there is still so much work to be done.

Evelyn and Savannah Rupert.

CRN Now Accepting 2024 Scholarship Applications

By Gail Potts, Director

Each year the CRN provides scholarship opportunities for students currently enrolled in, or preparing to begin an accredited collegiate or vocational program. Financial assistance of $2,000 USD will be awarded for each of the three established scholarships. Scholarships are available to an Individual Living with Cystinosis, the Sierra Woodward Sibling Scholarship and the Deanna Lynn Potts Scholarship.

The Sierra Woodward Scholarship was established in memory of Tahnie Woodward's sister. The Deanna Lynn Potts Scholarship was established by the Potts family in memory and in honor of their daughter who passed away from Cystinosis in 2000. It was Deanna Lynn's wish to establish this scholarship upon her passing. She wanted to see others with Cystinosis have some financial aid in furthering their career goals.

Since the Deanna Lynn Scholarship was established in 2000, there have been nineteen awards given to individuals with Cystinosis. The Individual Living with Cystinosis Scholarship was established in 2005 by CRN awarding nineteen scholarships. The Sierra Woodward Sibling Scholarship was also established in 2006 and has provided 20 awards.

The annual scholarship deadline is August 1. We are now accepting applications for 2024.

For more information, check our website at https://cystinosis.org/support-resources/scholarships/ or contact Gail Potts at gpotts47@yahoo.com.

Attention Students! Our scholarship amounts have doubled from $1,000 to $2,000.

Living with Cystinosis Support Group

A supportive, web-based
peer support group for those
living with cystinosis
facilitated by
Kerry Heckman, MSW, LICSW

3rd Tuesday of each month from
4-5pm PST / 7-8pm EST
To sign up, visit
thecenterforchronicillness.org/groups

Contact us at
info@thecenterforchronicillness.org
or (425) 296-2705 with questions.
www.thecenterforchronicillness.org
This program is free of cost.

Center *for* Chronic Illness

Supporting Loved Ones of those Living with Cystinosis Support Group

A web-based peer support group for loved ones of
those living with the rare disease, cystinosis
facilitated by Kerry Heckman, MSW, LICSW

3rd Tuesday of every other month
6-7pm PST / 9-10pm EST
(upcoming meetings: March 19th, May 21st, July 16th 2024)
To sign up, visit www.thecenterforchronicillness.org/groups

Contact us at info @thecenterforchronicillness.org
or (425) 296-2705 with questions
www.thecenterforchronicillness.org
This program is free of cost.

Center for Chronic Illness

Rare Disease Day at Quinnipiac University – Hamden, Connecticut

By Marybeth Krummenacker, Vice President of Education & Awareness

On February 20th, Laura Krummenacker, Emily Mello, Herberth, Jessica and Martina Sigler and myself were invited to present a rare disease to a group of students at Quinnipiac University by bringing in real patients. The opportunity was presented to us by CRN's Social Worker, Maya Doyle.

The students broke off into groups to discuss the patient history and draw their own conclusions to form a diagnosis. After their discussions, Laura, Emily and Martina were introduced as the real patients with this rare disease. Kristina Sevel joined us via Zoom and spoke to the students as well about the frustrations of a more recently diagnosed patient. She shared what she has learned in order to advocate for her daughter. The cheering section in attendance, their parents, watched in awe as they confidently answered the questions that were presented to them by Maya.

It was an interesting exchange to see these three young women answer whatever questions were given to them in an honest, calm and reflective way. It was powerful! The opportunity for the parents to give their input was also key to the discussions as they described obstacles that happened throughout their lives and how they overcame them. It was an extraordinary and wonderful opportunity to address the next generation of nurses, doctors and physical therapists in a frank and honest discussion. These three young ladies spoke eloquently and honestly about themselves, but made it very clear that just because they were diagnosed with cystinosis, didn't mean they couldn't do whatever they wanted to do.

It was a great day, and served as a reminder of how important it is to educate anyone who will listen about cystinosis. As I shared, 35 years ago in the ER, speaking to a young resident who said, "we learned about this disease in medical school, but the professor told us to not be concerned about it because you will probably never see it in your lifetime". I told the students to NEVER forget these three young ladies.

Rare Disease Day panel and guests at Quinnipiac University.

Laura Krummenacker, Emily Mello and Martina Sigler share their experiences with students at Quinnipiac University in honor of Rare Disease Day.

In preparation for our 2025 Family Conference at the beautiful Amway Grand Hotel, please check out their website (ExperienceGR.com) for some of the wonderful activities Grand Rapids, Michigan has to offer to make this a memorable event.

Help Support the CRN's Mission – Donate Today!

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

Become more active within our 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"

We are here to for you, whether you are:

- A parent who needs critical resource information, support services or help in sharing the challenges of cystinosis with those who serve your child
- An adult with cystinosis interested in information regarding medical and social issues that are specifically geared for adults or you are interested in contributing ideas to new and/or legacy programming through the Adult Leadership Advisory Board
- 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

The Cystinosis Research Network is proud to provide valuable resources to the community, free of charge. Many can be found on our website: cystinosis.org.

Resources include but are not limited to:

- The latest cystinosis information through our biannual CRN Newsletter; The Cystinosis Advocate, our website (cystinosis.org), the popular online cystinosis Facebook support groups, regular email updates and other social media channels.
- CRN Family Conferences and Regional Meet Ups. Exchange knowledge and create friendships with other families and individuals living with cystinosis. Learn first-hand the latest discoveries about cystinosis from medical professionals.
- Rare Disease Week Scholarships. Participate in a week-long event in Washington, D.C. Let your voice be heard by legislators and policymakers who need to know why cystinosis (and other rare diseases) is important to you.
- Access to Cystinosis Research Network individuals and families near you.

Please consider donating to the CRN today to help us continue offering robust support, education and research to the global community.

Ways to donate:

- Scan the QR code using your mobile camera
- Use the envelope provided to contribute via check
- Visit our website donation page at https://tinyurl.com/CystDonate

CRN Exhibits at the ASN Kidney Week 2023 in Philadelphia, Nov. 2-4

By Terri Schleuder, Director

Our theme for this year's CRN exhibiting opportunity was four-fold:

Advocacy
Sharing our personal stories, connections, and purpose.

Education
Giving out educational and support materials to those with an interest in learning more about Cystinosis.

Networking
Making closer connections with our industry partners, physicians, researchers, and other nonprofit organizations in attendance. We reconnected with 'old' friends and made 'new' ones.

Collaboration
Sharing ideas to improve everyone's experience by learning from each other.

One such opportunity was meeting Giovanni Valenti, from Italy, a presenter and researcher on one of four posters highlighting Cystinosis research at this year's poster exhibits. We all felt a strong bond as she kindly explained her research in lay terms. She listened to each of us explain our personal connections with Cystinosis, and then shared her thoughts which touched us deeply. I believe it speaks to the entire purpose of Kidney Week, and most especially rare disease.

"We are all connected. Your sorrow is our sorrow, and our hope is your hope." – Giovanni Valenti

Thanks to Carol and Garry Hughes, Gail Potts, Terri Schleuder and Clair Johnstone for putting in the time and effort for another successful exhibit experience at this year's ASN.

Gail Potts, Terri Schleuder, Garry Hughes, and Carol Hughes, exhibiting for CRN at the 2023 ASN in Philadelphia.

Carol Hughes & Gail Potts with Giovanna Valenti.

Gail Potts and Garry Hughes with new friends from the Dent Disease Foundation.

Carol Hughes, Dr. Roslyn Mannon and Dr. Julie Ingelfinger.

Carol Hughes with Dr. Joshua Zaritsky.

Development Update

By Jonathan Dicks, President and Vice President of Development

Finn, Theo and Ellie Dicks.

Dear Cystinosis Community,

As we step into the new year, I am overwhelmed with a deep sense of gratitude for all of you who continue to stand by the Cystinosis Research Network (CRN). Your steadfast commitment and relentless efforts form the bedrock of our progress, and it's a privilege to be part of this collective endeavor.

Looking back on the past year, our first in-person family conference since 2019 stands out as a beacon of unity and resilience in our memories. The lively atmosphere and heartfelt connections shared in Nashville, Tennessee, served as a powerful reminder of the strength within our community. Attendees from diverse backgrounds and experiences came together to share stories, offer support, and forge lasting connections, creating an atmosphere of solidarity and hope.

In particular, the conference witnessed an influx of newly diagnosed families seeking guidance on how to navigate their journey with cystinosis. Their presence underscored the importance of accessible resources and support networks, highlighting the growing need for initiatives that empower individuals and families facing this rare disease. As conversations unfolded and bonds were formed, it became evident that our community's collective determination to drive progress and foster mutual support knows no bounds.

With eyes fixed towards the horizon, I am deeply moved and elated (and a bit exhausted) to unveil another promising year ahead, with projected grant funding surpassing $500,000 for 2024! This significant backing stands as a tribute to the steadfast commitment and tireless contributions of our volunteers. Their unwavering dedication forms the bedrock upon which CRN's pioneering research and impactful initiatives thrive, propelling us towards our shared vision of a brighter future for all affected by cystinosis. Moreover, I must extend profound gratitude to our invaluable industry partners, whose generous support creates an unshakeable foundation, empowering us to advance our mission with unwavering confidence and determination.

Equally heartening is the expansion of our array of no-cost services, which continue to flourish. I am delighted to share the return of local CRN-driven in-person meet-ups, with the next gathering scheduled in Cincinnati, Ohio on May 4th! If you reside in the tri-state area (OH, IN, KY) or are within a 2-4 hour travel radius, access our event page to register for free today at tinyurl.com/4zww6dd3.

Furthermore, I'm honored to acknowledge the dedicated efforts of our devoted Conference Selection Committee, who have diligently coordinated our upcoming family conference. It fills me with immense pleasure to disclose that our chosen destination is the vibrant city of Grand Rapids, Michigan at the magnificent Amway Grand Hotel! This achievement would not be possible without the steadfast support of our industry sponsors: Recordati Rare Diseases, Amgen, Leadiant Biosciences, and Novartis. Their unwavering commitment underscores their belief in our mission and strengthens our resolve to make a difference.

Our dedication to serving the community remains unwavering. The Cystinosis Warriors Impact Program continues to inspire and empower, bringing together individuals and families affected by cystinosis. We take immense pride in the program's positive influence and its ongoing impact on the lives it touches.

In our ongoing collaboration with the PCs for People program, now in its third year, we reaffirm our dedication to accessibility and support. Notably, we're excited to announce a new development: alongside providing free computers and internet access to patients and caregivers within our community, we're extending this opportunity to accommodate a sibling as well. This initiative reflects our commitment to breaking down barriers and empowering individuals to flourish in their everyday endeavors.

Development Update, continued

We stand firm in our commitment to ensuring access to essential services for Spanish-speaking individuals and families in our community. Our dedication extends globally, now reaching cystinosis patients worldwide with translation solutions available in over 200 languages.

If you or a loved one has recently received a cystinosis diagnosis, or if you're navigating through the transplantation phase or transitioning to adult primary care, we're here to offer comprehensive support. Explore the resources provided by our Care Package Program, meticulously designed to assist individuals and families facing cystinosis during challenging times. Let us tailor a personalized package filled with informative materials and practical aid to suit your specific stage in the cystinosis journey. To register for your complimentary package, simply visit https://cystinosis.org/care-package/ and complete a brief form.

In closing, I want to express my deepest gratitude to each and every member of our remarkable community. Your unwavering support, dedication, and resilience continue to inspire us every day. Together, we have achieved so much, and I am filled with optimism as we journey forward into the new year. Let us continue to stand united, supporting one another, and advocating for those affected by cystinosis. With your ongoing support, we will overcome challenges, celebrate victories, and make meaningful progress towards our shared goal of a brighter future for all. Thank you for being an integral part of the Cystinosis Research Network.

With Love,
Jonathan Dicks

CNE International Conference 2024

By Will Newman, Chair of Organising Committee and Cystinosis Foundation UK

Better Together
CNE INTERNATIONAL CONFERENCE 2024
CYSTINOSIS FOUNDATION UK & CYSTINOSIS NETWORK EUROPE

Manchester UK
25 – 27 July

As hosts of the 2024 CNE International Conference we're really looking forward to welcoming you to Manchester in July. This is a team effort with Cystinosis Foundation UK and Cystinosis Network Europe working together to develop the very best experience. Better Together sums up our feelings about the strength of the cystinosis community. We know that families and individuals with cystinosis working in true partnership with clinicians, researchers and pharma can help accelerate improvements in care and treatments. And there is nothing better than being together to build friendships and connections that are the cornerstone of our community support. We have a fabulous venue, the Deansgate Hilton, with astonishing views over Manchester and easy access to all that this vibrant city can offer. So come and join us in July and experience being Better Together. You can find out more about the conference on the dedicated website page where you can also book your places, including accommodation (and travel guidance), via our simple registration portal. https://www.cystinosis-europe.eu/our-conference

The Cystinosis Memorial Fund Needs You!

By Karen Gledhill, Secretary

The CMF (Cystinosis Memorial Fund) needs you! The fund, which was established to help teens and young adults living with cystinosis, is waiting for your request. The goal of the fund is to instill added confidence, abilities and skills to its recipients to flourish and obtain their full potential.

The fund provides a stipend to help those living with cystinosis pay for a variety of added educational activities. These have included funding for a nursing course, professional workshops, certifications, career coaching, yoga workshops, nutritionist visits, resume writing, software programs, tutoring, and technology courses.

One of our most recent winners, Ashley Abedini, has shared her story.

“I own Abedini Social LLC, a one-woman social media marketing agency. I work with small businesses to manage their social media platforms, with a focus on Instagram, Facebook, and TikTok. I create on-brand content, engage with those in their target market, analyze social metrics, and utilize paid ads to make sure their social content gets a boost. I am passionate about using social media to help business owners find success, and I find joy in utilizing my marketing skills to help busy small business owners. Social media is paramount to so many businesses’ success, and I am grateful to have the opportunity to work for myself to help achieve this.

Ashley Abedini at the Wichita Chamvber of Business Expo.

Being a small business owner can be quite challenging at times, as well as costly. Small expenses often arise and easily add up. The CRN Memorial Fund has helped me offset some of the financial burdens of owning a business, including helping finance the printing for a local business expo and covering the costs of local business organizations. This assistance has allowed me to get more promotions within my local market and ultimately helped me attract more potential clients.”

Ashley Abedini, owner of Abedini Social, LLC.

Cystinosis MEMORIAL FUND

Learn more at cystinosis.org/cmf

WWW.CYSTINOSIS in FOCUS.com

INTRODUCING

A website with resources on eye care for people and their families living with cystinosis

Whether you have questions, seek insights into ocular symptoms of cystinosis, or want to hear from an eye care specialist, our website will provide practical information for patients and their families. The content is relevant for family members/caregivers of newly diagnosed patients, as well as teens and adults living with cystinosis.

WWW.CYSTINOSIS in FOCUS.com OFFERS MORE FOR YOU

- **A website** where you can learn about cystine crystal accumulation in your eyes
- **A website** where you can hear from eye care experts about what you may expect and how to support your eye health goals
- **A website** where you can download resources to help prepare for upcoming eye appointments

FOR MORE INFORMATION VISIT OUR WEBSITE AT

WWW.CYSTINOSIS in FOCUS.com

In Memoriam

John Sanders

Obituary published on Legacy.com by Williams-White Columns Funeral Home on Jan.12, 2024

Mr. John Alton Sanders, 58, of Ivey, passed away Sunday, January 7, 2024. Funeral services for Mr. John Sanders were conducted on Saturday, January 13, 2024 at Williams-White Columns Funeral Home at 2 o'clock with Rev. Danny Young officiating. Interment followed in Baptist Gospel Mission Cemetery in Twiggs County.

Mr. Sanders was a native of Bibb County but had made his home in Wilkinson County most of his life and was the son of Grady and Marie Sanders. John was an EMT and retired from BASF. He loved spending time with his family, fishing and riding his motorcycle. John had a passion for Benefit rides, which made him known for his kindness and huge heart. He was a straight talker and man of strong faith. John was a Veteran, having served in the Army and was a member of The Morris Little American Legion Post #6 in Milledgeville and was Legionair of the year 2023.

Survivors include his children; C.J. (Jessica) Sanders of Chester; Colin Sanders of Milledgeville and Camden Sanders of Ivey; his parents, Grady Alton Sanders and Marie Griffin Sanders of Gordon; brother, Rohn (Myrna) Sanders of Gordon; grandchildren: Eli, Harper, Jevan and Henry Sanders of Chester; nieces: Emily Sanders and Kaylee (Hunter) Hanson of Gordon and nephew:

John was a long time member of the cystinosis community and the father of cystinosis warrior, Camden.

Kannon Thomas of Gordon Williams-White Columns Funeral Home is in charge of arrangements.

Skyler Silvano Minella, 2003-2023

Obituary published on Legacy.com by Marana Mortuary & Cemetery on Nov. 29, 2023

It is with great sadness that we announce the unexpected passing of our beloved angel son Skyler Silvano Minella at the young age of 20. His entire life he battled Cystinosis and was a kidney donor recipient in 2018. He passed away with huge heart and a rainbow of light on Tuesday November 27, 2023 at Banner UMC.

Skyler Silvano Minella was born to the parents of Silvano Ralph Minella Jr and Christina Marie Minella. Skyler's life was full of family and friends that loved him. He was a ray of sunshine that could brighten any room and had a laugh that was infectious. His hobbies that he enjoyed was going to the movies, building Legos, gaming with friends, listening to music, and playing with his dogs.

He leaves behind his mother and father Christina and Silvano Minella, Jr.; two grandmothers, Sherry Jeppson and Monica Valenzuela; a grandfather, Chuck Jeppson; and predeceased grandfather, Silvano R. Minella, Sr. He will be deeply missed by all.

A funeral service was be held on Friday, December 1, 2023 at 11:00 am at Marana Mortuary and Cemetery with a visitation from 10:00 am until service time. In lieu of flowers please make donations to the Cystinosis Research Foundation https://www.cystinosisresearch.org/ or Cystinosis Research Network https://cystinosis.org/

In Memoriam

Janice B. Finn
June 29, 1983 – January 30, 2024

By Emily Mello

Janice Finn enjoying life near the water.

Emily Mello with her friend Janice Finn.

Both Janice, and I were in need of a friend when we met in 2017. Same rare disease, same transplant doctors, but mostly what connected us was the same love for food, musicals and Disney.

Janice was born June 29, 1983 to Nancy and Gerry Finn, in Hicksville, NY. Nancy and Gerald soon realized that Janice had something going on with her health and through mutual friends discovered that Janice had cystinosis. So began a new journey for the family with medications, doctors' appointments and all things related to rare disease. Even through diagnosis, medications, and the daily challenges of living with a rare disease, Janice never let any of those things stop her.

From Girl Scouts, meeting her life-long best friend Lori Kasprzak, to vacations, high school, college and a working adult life, SHE LIVED. Janice loved dogs, the beach and food.
We met at the exact, perfect time, through mutual healthcare providers who thought I could use a friend who also had cystinosis, and lived in CT. I agreed, and Janice agreed, and we got into contact with each other. Meeting Janice was a relief to me because she came into my life exactly when I needed a listening ear. It was friendship at first encounter. From then on, we would set our appointments on the same dates so we could hang out afterwards. Our go to hang outs were the cinema, to watch a movie and have Chic-Fil-a, concerts, the beach and trying out new places to eat! Our conversations and memories I will keep. She was the definition of a true friend. Janice was a daughter, a sister, an aunt, a true friend and an excellent hard worker. She was also a doggy momma to Chester and Snoopy. Janice will leave an empty space in many hearts and will truly be missed. A genuine, smart, kind and amazing person.

Family Support Update

By Chelsea Meschke, Vice President of Family Support

Happy Spring! What a wonderful time to start getting rid of those winter blues! Birds are chirping, and the sun is starting to shine here in Michigan! I wanted to take some time out and let you all know what is going on with Family Support here at the Cystinosis Research Network. We have our bi-annunal Family Meet-Up on May 4th in the beautiful city of Cincinnati, OH. The Cincinnati Marriott at River Center will be our gracious host. It will be a one day only event, starting at 8:30am and going until 4:30pm. Please register at cystinosis.org (under Events) to secure your spot! Hotel rooms are blocked for the weekend in case anyone would like to stay! We hope that everyone who attends walks away with a new sense of support, knowledge of self-care, and knowing more about our community and organization. One goal we want to achieve with this event is to explore how to strengthen support within our community,

There are a few definitions of support. One is, “Bear all or part of the weight of; hold up.” The second, “Provide with a home and the necessities of life.” These terms and these definitions are so profound in our community’s everyday life. As caregivers, we do all these things in excess. We hold up our children and our families in times of stress and times of difficulties. We bear the weight of the world on our shoulders and hold this weight up to keep everyone going. As caregivers, as fighters and warriors we look for opportunities to provide our home with all the necessities of life and a glimpse of “normal” if we can. As a caregiver, there is so much feeling and relation

Brian, Myles, Jaxon and Chelsea Meschke.

to that single word support. CRN maintains the goal of showing up for the definition of support to each and everyone of our community members. Please know that we are here to offer that support and care to any family member, parent, caregiver, and/or warrior that needs it. Thank you to the Cystinosis Family for showing your support!

We can’t wait to see everyone in Cincinnati!

Please email me for support needs or questions at chelseam@cystinosis.org.

Please check out our care package program at https://cystinosis.org/care-package or email Jen Wyman jwyman@cystinosis.org.

HAVE YOU ATTENDED AN IMPACT PROGRAM YET?

The IMPACT Program is specifically designed for people living with cystinosis and their families, and sponsored by Horizon Therapeutics throughout 2022

IMPACT

IMPACT Adults

IMPACT Teens

IMPACT Family & Caregivers

- **Learn about living with cystinosis**
- **Connect with others impacted by this condition—hear and share ideas, feelings, and helpful advice**
- **Learn about medication:**
  - the critical importance of continuous cystine control through cystine-depleting therapy (CDT)
  - PROCYSBI (cysteamine bitartrate) delayed-release capsules and delayed-release oral granules for the treatment of nephropathic cystinosis
- **Meetings will occur in both the virtual (online) setting and live, in person**

Even if you've attended an IMPACT Program previously, please check back in with us!

The IMPACT Program has new topics created specifically for different cystinosis patient experiences—**adults, teens, and families/caregivers** of people living with cystinosis.

To RSVP to one of these meetings or to find out about programs occurring near you, please call 602-953-2552 or visit www.procysbi.com/Cost-Savings-and-Support

USE AND IMPORTANT SAFETY INFORMATION

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. **Tell your doctor right away if you get a skin rash.** Your dose of PROCYSBI may need to be decreased until the rash goes away. If the rash is severe, your doctor may tell you to stop taking PROCYSBI.

PROCYSBI®
(cysteamine bitartrate)
delayed-release capsules
delayed-release oral granules

Please see additional IMPORTANT SAFETY INFORMATION on the next page, and visit www.hzndocs.com/PROCYSBI-Patient-Information.pdf for the Patient Package Insert.

IMPORTANT SAFETY INFORMATION (continued)

- **Stomach and bowel (intestinal) problems.** Some people who take other medicines that contain cysteamine bitartrate may develop ulcers and bleeding in their stomach or bowel. People treated with PROCYSBI may also develop abnormal swelling and narrowing of the large bowel which must be treated promptly. **Tell your doctor right away** if you get abdominal pain, bloody or persistent diarrhea, bloating, nausea, vomiting, loss of appetite, vomit blood, poor weight gain or weight loss.
- **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 is PROCYSBI?

PROCYSBI (cysteamine bitartrate) delayed-release capsules and delayed-release oral granules is a prescription medicine used to treat 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.

Do not take PROCYSBI if you are allergic to penicillamine or cysteamine.

Before taking PROCYSBI, tell your doctor about all your 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.

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

What should I avoid while taking PROCYSBI?

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

What are the possible side effects of PROCYSBI?

PROCYSBI can cause serious side effects, including:

- **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, pink eye, diarrhea, cold, tiredness, flu, headache, problems with body salts or electrolytes, infection of ear, nose or throat, joint pain.

These are not all the possible side effects of PROCYSBI. Call your doctor for medical information about side effects. You may report side effects to FDA at 1-800-FDA-1088.

For additional important safety information, please visit www.hzndocs.com/PROCYSBI-Patient-Information.pdf for the Patient Package Insert, and discuss with your doctor.

PROCYSBI®
(cysteamine bitartrate)
delayed-release capsules
delayed-release oral granules

HORIZON

Good News

With this issue, we are beginning a new Feature in our Cystinosis Advocate Newsletter called ‘Good News’. In it we would like to celebrate and recognize life’s beautiful moments among our Cystinosis Warrior Community. Our hope is, folks would send us a photo with a caption highlighting memorable events that have occurred over the year, such as weddings, graduations, new babies, new jobs, successful transplants, or anything else notable to celebrate.

Today, we are honoring Dylan and Shannon (Keizer) Henderson who were married a few weeks ago in Kalamazoo, Michigan, and Heidi Hughes, who competed in the World Transplant Winter Games 2024 in Bormio, Italy in March.

If you would like to share ‘Good News’ in future editions please email a photo and a detailed caption or small paragraph to Terri Schleuder describing the event to: tschleuder@yahoo.com.

“Dylan and I are honored to share the good news of our recent wedding and marriage! Many of you had the privilege of meeting him this past summer at the conference in Nashville. He and I shared our first slow dance at the Saturday evening dinner dance, and we used the same song for the first dance at our wedding. Thank you for welcoming Dylan into the Cystinosis family with open arms!”

- Shannon Henderson

Heidi Hughes in the Italian Alps about to embark on a 5k cross country ski race for the 12th Annual World Transplant Winter Games in Bormio, Italy.

“I will always reach for the top of all my metaphorical and real life mountains. Cystinosis will not hold me back.”

- Heidi Hughes

Jovi's Story

By Raven Russell

I remember the day I met Jovi. The placement coordinator for our foster care agency called us around 10:30 that morning and shared details about a sweet baby girl in the hospital, who had already endured much suffering in her 7 weeks of life. Though we weren't really planning on bringing another little one into our home right then, we knew we wanted to show her all the love and care we had to give, while we helped her to heal physically and emotionally. The coordinator gave me all the necessary information and I was at the hospital before noon to meet with her medical team, and learn what we needed to know in order to bring her home.

Jovi Russell.

Raven holding Jovi and Matt holding Whit.

Though Jovi's body healed much over the next several weeks, learning to trust and open herself up to bonding was another story entirely. Over time, though, she became ours and we became hers. Seeing her form attachments was an answered prayer. It was such a special time in our family. She blessed us, and we all are forever changed because of her. Another level of 'precious', though, was her bond to our little guy, who we were fostering at the time. He is one month older than her. Whitman loved to be near her, and she was clearly comfortable with him. Whit would even instinctively reach out and hold her hand when she cried, causing her to calm. It was clear that this relationship, too, was a part of God's plan.

Whit and Jovi Russell.

Jovi had many medical appointments, and because of some things the medical care team noticed when she came into care, Jovi was to see a geneticist. In late spring, we were at last able to be seen. Genetic testing was ordered, and the results came in when Jovi was around 10 months old.

I received a call saying that, through testing, the doctor was able to rule out what he was looking for, and that a prior diagnosis would stick. When I thought the call would be coming to an end, I was told that there was actually more we needed to discuss. The testing revealed something they weren't looking for; an issue with her CTNS gene. We would have an appointment in one week's time, but in the meantime, I was told I should get online and research Cystinosis. I was not prepared for what I found. I guess, because I was told via phone, I didn't think it would be something too serious. I was mistaken. I shared all I knew and read with my husband,

Jovi's Story, continued

Jovi resting.

and we certainly cycled through a few emotions. Each night I would read for hours; every article I could find.

The week went by, and our appointment came. Our geneticist and a couple of his colleagues were there. One whom he asked to attend, was very knowledgeable about Cystinosis and explained things in detail. He first asked me to explain what I knew and then filled in the gaps. The next morning we had labs drawn. When the results came in, we received our referral to nephrology. Because Jovi clearly would not be able to take her medicine by mouth, she received her g-button a few weeks later. Then we waited several weeks for her medicine. It was a stressful process, and took many phone calls and much coordination on my part.

I never dreamed of the level of advocating this would take, but through it all, I have definitely sharpened my skills! We are early on in this diagnosis, and still figuring things out, and adjusting meds. Her levels are not yet under control, but for the first time, I feel like we are on a good, consistent, and purposeful course.

Along the way of becoming a parent of a child with cystinosis, I have taken special notice and want to share things that have happened or caught my attention:

1. We wrestled with many emotions in the beginning, but God has now given us an amazing peace beyond what we could have imagined.

2. We have felt lonely in this diagnosis, but the cystinosis community has stepped in, and been incredible. I am convinced this group is just built different, and I'm here for it! You treated me like an old friend even when you knew little more than my name.

3. We are learning more every day, and I can see that there will always be much to learn, especially since there is new research and new findings all the time. I'm so appreciative that Jovi's cystinosis was caught early through genetic testing.

4. We are very thankful that we get to walk with Jovi through her illness. Of course, we don't want this for her. But we love her so much and would not want her to experience this without us being here to care for her, comfort her, and fight for her!

Oh! Lastly, I need to update you on our Jovi's foster care status. Matt (my husband) and I didn't plan for, or know adopting would be a part of our fostering journey. Many children have come through our home. Amazingly, not one, but two children, were placed in our care within a few months of one another who would need a family. While we find it a bit amusing to be starting over in our parenting journey, we would not have it any other way. Watching our older children with the babies, have touched our hearts deeply. I could not imagine something more beautiful. It is extremely clear to all of us that Whit and Jovi are meant to be ours. I'm happy to report that Whit's adoption was final over a month ago, and by the time you read this newsletter, Jovi's adoption day will have come and gone too!

Matt reading to Jovi.

The Russell Family.

American Kidney Fund®
FIGHTING ON ALL FRONTS

NATIONAL VIRTUAL
AMERICAN KIDNEY FUND
CAMP

Join us for NATIONAL VIRTUAL Cystinosis Camp!

Virtual camp enriches the lives of children with kidney disease by allowing them to just be kids and connect with others who share similar experiences. Share life experiences, form friendships, socialize with peers and engage in creative, constructive virtual activities led by dedicated AKF staff.

Learn more and register today at www.kidneyfund.org/national-virtual-cystinosis-camp

Interact with other kids with cystinosis from across the nation!

Participate in dedicated virtual activities like art classes, games, science experiments & more!

100% virtual so you can log on from wherever you are!

Questions? Contact Lianna Chase at lchase@kidneyfund.org

Generously Sponsored By:
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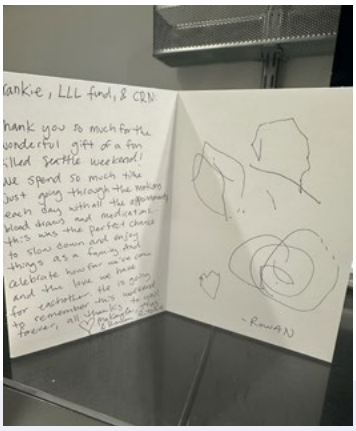
Live Like Laura Fun Fund: Recipient Rowan Ritchie and Family

By Makayla Sample



A huge thank you to the CRN and the Live Like Laura Fun Fund!! We had such an amazing time staying the weekend in Seattle! We ate lots of yummy food, went to the Seattle aquarium, the great wheel, the carousel downtown by the waterfront and even popped over to Bellevue for the snowflake lane parade. Followed by dinner at the Cheesecake Factory! Our day to day is so hectic and full of medications and doctor's visits, so this felt like the perfect opportunity to just have fun as a family.

The Live Like Laura Fun Fund was established in memory of Laura McGinnis, to allow those who live with cystinosis to experience life's adventures and fun as Laura loved to do. Learn more about the fund and how to apply at: cystinosis.org/lllff

Dutch Cystinosis Support Foundation - Research Updates

By Marjolein Bos and Fons Sondag

At the RadboudUMC in Nijmegen the Netherlands Annemarie de Vreugd just started on a PhD research project called 'Improving Patient Oriented Care in Cystinosis'. This research is sponsored by the Dutch Cystinosis Support Foundation. The main topics are:

1. Patients reported experiences and comparison of patients treated by IR cysteamine and DR cysteamine

2. Physical, occupational and speech therapy in cystinosis; where should attention lay, and which advice is valuable in daily life?

3. Course of Cystinosis above the age of 40. What are the symptoms and problems people face after 40?

4. Gastrointestinal complaints in cystinosis; evaluation of the GI problems, and effect on daily life and body composition

5. Cystinosis patient registry; evaluation of the European cystinosis population

6. Psychosocial burden in patients with cystinosis and the effect on QoL and self-efficacy

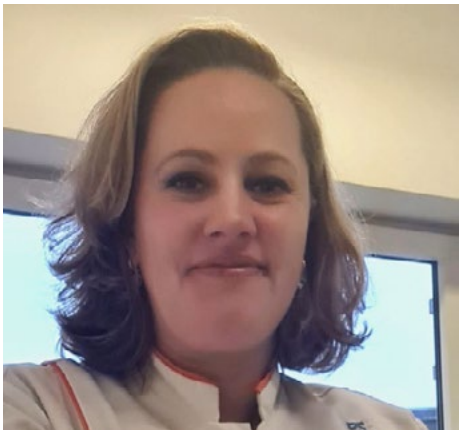
The most important tools in this research will be inquiries, interviews with patients and registry data. Annemarie expects to finalize this research at the end of 2027. If you would like to get in touch with Annemarie about this research, please mail her at: annemarie.devreugd@radboudumc.nl.

Email: cystinose@ziggo.nl

Website: www.cystinose.nl

CYSTINOSE Groep

Nederland en Vlaanderen



Annemarie de Vreugd.

10th Annual Dublin Cystinosis Workshop

By Denise Dunn

Attendees of the 2024 Dublin Cystinosis Workshop.

2024 Roz Anderson Prize awarded at the Dublin Cystinosis Workshop.

In February, Cystinosis Ireland invited 40 experts in cystinosis research and treatment, as well as, experts working in the field of myopathy (muscle wasting) to join us for the 10th annual Dublin Cystinosis Workshop. Speakers included keynote addresses from Dr. Reza Sadjadi, Harvard Medical School, USA, on clinical trial readiness, myopathy and dysphagia assessment in adults with Nephropathic Cystinosis; and Philippos Mourikis of East-Paris University, France, on Muscle stem cells, and their niche and how it can be used to study severe and mild myopathy. Projects currently funded by Cystinosis Ireland were presented in video updates.

The winners of the 2023 Professor Roz Anderson memorial prize, and the Cystinosis Ireland award for the best oral presentation to a non-scientific audience, Louise Medaer and Hayley Chang respectively, also shared updates on the progress in their work. The meeting was delighted to award the 2024 Professor Roz Anderson memorial prize to Sante Princiero Berlingerio of KU Leuven, Belgium presenting his work “Targeting oxidative stress-driven lipid peroxidation improves podocyte dysfunction in cystinosis”. You can find out more about the Dublin Cystinosis Workshop at www.cystinosis.ie/research.

Cystinosis Ireland continues to support the work of Cystinosis Network Europe (CNE), the global network of national organisations which support people living with cystinosis with research, and promotes research into the condition. The CNE international conference will be hosted by Cystinosis Foundation UK in Manchester in July and more details can be found on www.cystinosis-europe.eu/our-conference. CNE also works with promoters and sponsors of research at all stages in its development through our Community Advisory Board. More details on its work can be found at www.cystinosis-europe.eu/community-advisory-board.

Cystinosis Mexico Updates

By Victor Gomez

Recently our organization has formed its 2024-2025 Board of Directors, a group of parents/patients committed to the guidelines of our organization, by supporting patients with Cystinosis in Mexico.

Among the lines of action for this year we will work on:

- Establishing the cystine level analysis center in Mexico
- Training of medical specialists
- Promoting a special attention system for patients with Cystinosis in Mexico supported by the health system, and supported by the government

Financial Update

By Tim Wyman, Treasurer

The Cystinosis Research Network continues to utilize its financial resources to further its mission 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. Additionally, since 1996 CRN's vision includes enhancing the quality of life for those with cystinosis. To that end, CRN expended significant resources in 2023 for the Nashville Family Conference which brought together hundreds in the cystinosis community along with both medical and industry partners.

Total income for 2023 was $510,966 which consisted of over $58k in fundraising efforts and $436k in grants received from our industry partners. Both of these numbers were higher than 2022 which has been consistent with conference years. As expected, expenses were higher in a conference year and totaled $694k for -$183k in net income. As noted, both CRN's income and expenses in a conference year are significantly higher than non-conference years and the majority of expenses were attributable to the conference and scholarships. CRN's current equity (assets minus liability as of December 31, 2023) stands at roughly $273,800 which is critical in funding additional research.

The CRN is a tax-exempt organization granted "501(c)(3)" nonprofit status by the I.R.S. The CRN Federal Tax ID # is 04-3323789.

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What is CYSTARAN®?

CYSTARAN (cysteamine ophthalmic solution) 0.44% is an eyedrop medication used to treat cystine crystal accumulation in the corneas of patients who have cystinosis

What is the most important information I should know about CYSTARAN?

- To help prevent contamination of the dropper tip and eyedrop medication, try to make sure that CYSTARAN is dropped directly onto the eye without touching it. Try not to touch the eyelids or surrounding areas with the dropper tip of the bottle when you are using CYSTARAN. Keep the bottle tightly closed when not in use.
- CYSTARAN contains an ingredient called benzalkonium chloride which can be absorbed by soft contact lenses. Remove contact lenses before using CYSTARAN eyedrops and wait at least 15 minutes before reinserting them.
- CYSTARAN should only be used as an eyedrop medication.

What are the side effects of CYSTARAN?

- The most common side effects of CYSTARAN, which have occurred in at least 10% of people using the medication, were sensitivity to light, eye redness, eye pain and irritation, and headache.

The risk information provided here is not comprehensive. To learn more, talk to your healthcare provider or pharmacist about CYSTARAN. The full FDA-approved product labeling can be found at www.cystaran.com.

You are encouraged to report negative side effects of prescription drugs to the FDA. Visit www.fda.gov/medwatch or call FDA at 1-800-FDA-1088.

Scan the QR code to learn more about CYSTARAN
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For information about ordering CYSTARAN®, please contact our partner, AllianceRx Walgreens Pharmacy:

PHONE: 1-877-534-9627

Research Update

By Kristina Sevel, Vice President of Research

As we delve into the latest edition of our Cystinosis Advocate newsletter, we are proud to share continued updates on our ongoing research initiatives. I am continuously awed by the unwavering dedication and passion displayed by this community. The efforts and commitment to advancing cystinosis research are the driving force behind our collective progress and success. Together, we are supporting new treatments, pushing boundaries, and working to make a tangible difference in the lives of those affected by cystinosis. As we fly in to 2024 my goal for the future is to expand our network of partnerships with leading researchers, healthcare professionals, and industry stakeholders to leverage collective expertise and resources. By fostering collaboration, we can maximize the impact of our research efforts.

To date CRN has funded over $5.5 million total in research grants and fellowships, including 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, Germany and much more. CRN has also co-funded research projects with Cystinosis Ireland and does so currently. CRN research topics have focused on every aspect of cystinosis with the purpose of understanding the disease and finding improved treatments and a cure. Topics include research and therapies related to neurological, genetic, ophthalmological, gastrointestinal, muscular, nephrology, pulmonary, skin, fertility, improved medications, psychological and much more. It's our honor to collaborate with our international cystinosis advocacy colleagues to support the best researchers around the world.

Current CRN Grant Commitments

Development of a patient-reported outcome to measure the health-related quality of life of children and adolescents with cystinosis

Drs. Katharina Hohenfellner and Julia Quitmann

Grant Amount: $155, 075.09, two year study

Patient-Reported Outcome measures (PROM) are questionnaire-based tools that can help healthcare professionals understand the health status or disease burden from the patient's perspective. These tools can be used to evaluate new therapies or to improve the healthcare provided. Disease-specific instruments that measure health-related quality of life (HrQoL) is particularly informative, as they capture the needs and challenges of specific patient groups particularly well. As a multidimensional construct, HrQoL includes physical, emotional, mental, social, and behavioral components of well-being from the patient's perspective HrQoL can be measured using four different types of instruments: generic, chronic-generic, condition[1]specific and treatment-specific instruments. Generic questionnaires represent the full range of health conditions, address groups independent of their respective health state and are effective for comparisons between two cohorts (e.g., patients with cystinosis and healthy controls). Chronic-generic instruments are focusing on a chronic condition independent of its specific characteristics, while specific questionnaires are tailored to problems associated with a specific condition (e.g., cystinosis) or treatment (e.g. patients receiving a kidney transplantation). Despite the significant impairments experienced

Grace and Kristina Sevel.

by patients with cystinosis, very few studies investigate HrQoL in this patient group and e disease specific HrQoL measures are lacking. Thus, the primary aim of this planned study is to develop a PROM for children and adolescents with cystinosis. This instrument will capture the HrQoL from both the child/adolescent and parent perspectives. It will be applicable to clinical trials ranging from randomized clinical trials (RCTs) to surveillance designs, focusing on the impact of cystinosis and its treatment. The preparations have already started. We are currently developing the questionnaire "QUALIFY" (Health-related quality of life of children and adolescents with cystinosis) through intensive literature research and interviews with young German patients and their parents. This preliminary version of QUALIFY needs to be cross-culturally validated in a larger sample (= investigated whether the instrument measures what it is supposed to measure) and adapted to the English, Spanish and French language. The study will

take place in four phases within a 24-month timeline (Figure 1). Patients and parents will be recruited by both clinicians and patient organizations of the participating countries. In the first phase, the preliminary German version of QUALIFY will be translated into English, French and Spanish. In phase 2, these new versions of QUALIFY will be used in an online pilot test to analyse their preliminary psychometric properties. A cognitive debriefing will be conducted so that patients and parents can reflect on the comprehensibility, completeness, and applicability of the instrument. In the third phase, the refined questionnaires will be applied in an online field test to evaluate their final psychometric properties (incl. internal consistency, convergent validity, reliability). In the fourth phase, a final report and publications for scientific journals will be written. The final product will be a cross-cultural, psychometrically validated, practically feasible, and conceptually suitable instrument for children and adolescents with cystinosis. It will be available in English, Spanish and French for further (inter-)national studies. With further international collaborations, this tool can be linguistically validated and cross-culturally 2 adapted for use in a wide range of countries. Although the project in this grant application refers to the development of PROM for children, the development of a PROM for adult patients is planned as a follow-up project.

Cognitive Control Systems in Cystinosis

Sophie Molholm, PhD Co-Principal Investigator, John Foxe, PhD Co-Principal Investigator

Grant Amount: $315,193, two year study

Seminal behavioral studies by Trauner and others in human patients have suggested the presence of cognitive dysfunction in cystinosis. Our own work suggests some behavioral and neural differences and difficulties with sensory memory in this population. However, the neurocognitive phenotype associated with CTNS mutations and its developmental path are still poorly understood, and the prime areas of neurocognitive vulnerability in this population are in need of much more thorough characterization. This is critical to developing effective therapies to compensate for or improve on areas of cognitive vulnerability. To this end, we propose to characterize different components of executive functioning (memory updating, set shifting, conflict monitoring, and inhibition) in cystinosis. Focus on this area is motivated by our previous work, the literature on cognitive weaknesses in cystinosis thus far, and by first person reports collected during interactions with patients and families. We will use high-density electrophysiology (EEG)—a non-invasive method that allows one to directly measure functional brain activity at the millisecond scale and thus reliably assess the integrity of information processing at the neural level—and standardized cognitive functional assessments to test 15 children, 15 adolescents, and 15 adults with cystinosis and the same number of age-matched unaffected healthy controls. Twenty heterozygotes and age matched controls will also be tested to examine the role that mutation versus disease plays in the cognitive phenotype of cystinosis,

Significance: Greater knowledge of the neurocognitive dynamics of executive function in cystinosis has the potential to critically advance the development of interventions for these individuals. In addition, identification of neural markers will provide objective assays of treatment efficacy on brain function.

Chitotriosidase as a Therapeutic Monitor for Cysteamine Therapy in Cystinosis: a Retrospective Validation Study

Mohamed A. Elmonem, Koenraad R.P. Veys, Lambertus P. van den Heuvel, William A. Gahl, Elena Levtchenko

Grant amount: $44,000, one year study

Nephropathic cystinosis lacks a practical therapeutic monitor for cysteamine therapy. The currently used white blood cells (WBC) cystine assay, although very specific for the disease, has many clinical and technical drawbacks limiting its routine use in most countries. Chitotriosidase enzyme activity in plasma is a potential alternative to WBC cystine in nephropathic cystinosis monitoring. Chitotriosidase is mainly produced in various tissues by activated macrophages upon stimulation by cystine crystals. We have previously demonstrated that the enzyme is significantly elevated in the plasma of cystinosis patients compared to both healthy and renal controls. Moreover, in a two-year longitudinal study chitotriosidase enzyme activity was a significant independent predictor of WBC cystine levels and was superior to WBC cystine as an indicator of the number of extrarenal complications in cystinosis patients. When compared to WBC cystine, plasma chitotriosidase assay is simpler, faster, more economical and needs a much smaller sample making it more convenient, especially in young children. Furthermore, the enzyme is extremely stable for years at minus zero temperatures. In the current proposal, we plan to validate our results in a large cohort of previously recruited cystinosis patients followed up at the NIH Clinical Center in Bethesda, Maryland, USA. Stored samples at -80°C of 150 cystinosis

Research Update, continued

Mike, Kristina and Grace Sevel.

patients will be retrospectively assayed for chitotriosidase enzyme activity, and will be correlated to documented parameters of kidney function, extra renal complications and a WBC cystine-based long-term compliance score.

IMPACT – Improvement of Motoric Abilities in Patients with Cystinosis

Katharina Hohenfellner, MD

Grant Amount: $77,000, one year study

The concept presented here, follows the established approach for the rehabilitation of children “Auf die Beine”, which was developed in Cologne for children and adolescents with limited mobility, e.g. caused by cerebral palsy or patients with osteogenesis imperfecta (https://unireha.uk-koeln.de/kinder-jugendreha/behandlungskonzept-auf-die-beine/). The training design with home exercise and short training sessions considers the fact that the target patient group is already under great strain due to the severity of the disease (medication, special diet, possibly dialysis). The project follows the official recommendations for the use of a vibration plate (4,5). Patients train with Galileo vibration plates according to a fixed training schedule which provides for 10 short training sessions per week (maximum 2 per day). Within one training session, four exercises will be performed. The control group will perform the same exercises without vibration plates, but with dumbbells. Patients will initially undergo an intensive training course and will receive regular supervision during the three-month home training phase. The study is designed as a randomized controlled trial. Due to the very rare underlying disease a matched pair design was chosen to achieve comparability between the two groups. The patients will be matched based on their age, sex, and major previous surgeries. A baseline and two follow-up clinical assessment, one after the three-month home training phase and one after the follow-up phase, will take place. The primary endpoint is the change in muscle strength (in %) from the baseline examination to the measurement after the training phase. In the control group (CG), a mean shift of 0 % is expected, since no vibration training is carried out. Patient orientation: empowering patients by supporting an active lifestyle and enabling patients to positively influence the course of the disease themselves. Optimization of clinical outcomes: improving cardiorespiratory performance and increasing muscle strength in patients. Improvement of patient-oriented end points of care: improving quality of life.

Grant Awarded February 2021 by the Cystinosis Research Network and Cystinosis Ireland

Perturbations in the V-ATPase Pathway Drive Pathology in the Male Reproductive System in Cystinosis

Principal Investigator Professor Minnie Sarwal, Professor of Surgery, Division of Multi Organ Transplantation, University of California San Francisco (UCSF), USA and co-applicants, Dr James F. Smith, Associate Professor and Director Male Reproductive Health, Department of Urology, University of California, San Francisco and Dr Polina V Lishko, Associate Professor, Department of Molecular and Cell Biology, University of California Berkeley, USA

The research project is a total investment of €300,000 from Cystinosis Ireland and CRN (€150,000 each) over the next three years.

Cystinosis is a very rare inherited genetic disease that causes the build-up of cystine, an amino acid is normally present in very small amounts in every single cell of a healthy person. The excess cystine forms sharp crystals that damage the body’s cells. Many of the body’s organs are affected by cystinosis including the kidneys and the eyes in particular. However in men, there can be an impact on fertility and the ability to produce sperm (azoospermia). Whereas in the past, the life expectancy of men living with cystinosis was short and their physical wellbeing relatively poor, today there are an increasing proportion of men living with cystinosis who are well and who want to consider parenthood. This research project aims to study the molecular and cellular changes that can cause azoospermia in men with cystinosis. The research will be a first step towards developing an effective treatment that will give men living with cystinosis the opportunity to become fathers. The knowledge generated from this research will also improve our overall understanding of the disease and in particular of certain poorly understood cystinosis symptoms that appear to be caused by malfunctions other than the accumulation of cystine. In selecting

this proposal for co-funding, the Boards of Cystinosis Ireland and CRN agreed that this is a scientifically significant proposal focused on a very important and strategic research topic for cystinosis patients. This project builds upon research and results generated from two previous projects co-funded by Cystinosis Ireland and the Irish Government's health research funding agency (the HRB) – a project led by Professor Minnie Sarwal in UCSF, USA entitled "Targeting Autophagy in Nephropathic Cystinosis" and a project led by Professor Elena Levtchenko in UZ Leuven, Belgium entitled "Unravelling the mechanisms of azoospermia and potential future treatments in male cystinosis patients".

CRN and Cystinosis Ireland Co-Fund UCSF Study of Male Infertility

We are pleased to announce a collaboration between the Cystinosis Research Network (CRN) and Cystinosis Ireland to fund a male infertility in cystinosis study at the University of California San Francisco (UCSF). The collaboration was made possible, in part, from a private contribution from CRN board members, Anna and Paul Pruitt and from Cystinosis Ireland's Seedcorn Funding Programme. Dr. Sur, a postdoctoral fellow in Professor Minnie Sarwal's Laboratory at UCSF, is the Principal Investigator in the "Cellular Resource for Studying Male Infertility in Cystinosis" proposal. Both organizations look forward to this partnership and providing greater insights concerning cystinosis and male infertility.

A Cellular Resource for Studying Male Infertility in Cystinosis, Minnie Sarwal, MD, PhD, Professor of Surgery, (Director, Precision Transplant Medicine) University of California San Francisco (UCSF), James Smith, MD, MS (Director, Male Reproductive Health Center Urologist), University of California San Francisco (UCSF), Ann Harris, Professor, Department of Genetics and Genome Sciences, School of Medicine, Case Western Reserve University, Cleveland, Ohio, Elena Levchenko, Professor, Department of Pediatric Nephrology, Leuven, EU, and Swastika Sur, MSc., PhD Postdoctoral Scholar, Sarwal Lab, University of California San Francisco, Department of Surgery.

Total Grant: €10,000

Principal Investigator, Swastika Sur, a Postdoctoral Fellow at the University of California San Francisco, has outlined a research proposal focused on infertility in men with cystinosis.

In addition to various endocrine organs that are affected in cystinosis, hypergonadotropic hypogonadism has been reported as a frequent finding in male cystinosis patients. Although spermatogenesis has shown to be intact at the testicular level in some patients, no male cystinosis patient is known to have naturally fathered a child. The sole treatment for cystinosis is the aminothiol cysteamine, which is highly effective in reducing the intracellular levels of cystine. However, this drug is ineffective in the treatment of male infertility in these patients.

A previous study of the pathophysiology of cystinosis-mediated infertility used a CTNS-/- mouse model. However, the CTNS-/- mouse model generated on C57BL/6 background was found to not be suitable for clarifying the pathogenesis of male infertility in cystinosis. This mouse model partially mimics the renal phenotype of the human disease but was found to have normal testicular morphology and function.

Therefore to this date, the exact pathophysiology of male infertility observed in patients with cystinosis is not yet fully understood, which is a critical unmet need due to the growing population of cystinosis patients, treated with cysteamine reaching young adulthood.

This project proposes to generate isogenic immortalized epididymis and testicular cell models to study infertility associated with cystinosis, by using CRISPR/Cas9 technology to develop a deletion in the CTNS gene of normal human epididymis and testicular cells. The Sarwal group's ongoing collaborations with Dr Smith at UCSF has enabled access to a rich tissue resource of normal testicular and epidydymal samples that will be used for generating this cystinosis- specific resource. The Sarwal research group also has ethical approvals in place at UCSF and at KU Leuven that will allow Dr Sur to approach cystinotic patients for access to patient biosamples.

Previously, the Sarwal research group successfully generated human immortalized CTNS-/- proximal tubular epithelial cells, and confirmed the phenotype of these newly developed cell line in terms of intracellular cystine levels by HPLC-MS/MS.

In this project, Dr Sur will focus on generating human immortalized CTNS-/- epididymal and testicular cells, followed by phenotype validation so that these cell lines can serve as a resource for the research community to study the pathophysiology of male infertility observed in cystinosis.

The Specific Aims of this project are the following:

· Aim 1: Generate human immortalized CTNS -/- epididymal and testis cell lines by CRISPR/Cas9 and confirm the phenotype to further downstream study of male fertility associated with cystinosis

· Aim 2: Map the molecular perturbations in both cell lines with

deletion of CTNS and in tissue samples from male cystinotic patients, by using state of the art genomics that the Sarwal Lab has legacy expertise-in. This will define the clinical utility of the resource generated in Aim 1.

This project will provide a first of its kind human cellular models to study cystinosis-mediated male infertility.

Cystinosis Community Advisory Board/Cystinosis Network Europe

I look forward to taking on the position as the U.S. representative in the Cystinosis 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. We look forward to continued partnership with researchers and industry worldwide to improve the quality and speed with which Cystinosis treatments are developed with the patient's voice in mind.

National Institutes of Health

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

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

Educational Resources

All of CRN's educational materials including brochures, guides and other publications have been updated and are available on the CRN Website. Look for an expanded Dialysis and Transplant section coming soon which will include a broad range of information and resources for those facing these challenges.

Please visit the Research page on the CRN website for updates on CRN funded studies as well as other research from the world. Also be sure to check out the many cystinosis related articles and publications available in our Publications and Guides library at https://www.cystinosis.org/support-resources/publications-guides/

Impact of accelerating genomic diversity on rare diseases

by Neveen A. Soliman, MD, PhD

Professor of Pediatrics, Kasr Al Ainy Medical School, Cairo University

Chair of Egypt Genome Scientific Committee

Genomics is the study of human complete gene code and how the structure and function of our genome relates to health and disease. Understanding the genetic basis of the rare diseases informs diagnostic and therapeutic healthcare strategies as well as predicting rare diseases.

The genomic revolution over the past two decades not only resulted in significant advances in rare disease research but also propelled the establishment of nationwide large-scale genome projects aiming to integrate genomics into mainstream healthcare. An essential component of many of such genome projects is dedicated to studying rare disease.

The impetus for the rare disease genomics in national genome projects, particularly in highly consanguineous populations, comes at a time when research into rare diseases is witnessing huge momentum. It also coincides with the emergence of rare diseases as global public health priority as identified by the World Health Organization within the vision of the Sustainable Development Goals "no one is left behind".

Questions that comes to mind are: what is genomic diversity? and how can it advance our

understanding of rare genetic diseases beyond the currently available knowledge?

Firstly, genomic diversity refers to the differences in DNA sequence between individuals and populations. While humans share about 99.8% of their DNA, the remaining 0.2% differ between individuals and if altered can have a significant impact on an individual's health. By studying the genetic variations that exist within and between populations, scientists can better understand the genetic basis of rare disorders and develop new treatments. After the mapping of the human genome in 2003, scientists realized that the potential benefit of genomic research towards precision medicine is limited by lack of genomic diversity with underrepresentation of other populations of African and Asian descent.

Secondly, studying how rare disease genetic variations fare within and between populations particularly where endogamy is common is crucial. Promising genome programs include the Middle Eastern programs in Qatar, Saudi Arabia and Emirates and in Africa the Egypt Genome Program that is majorly focused on rare disease genomics.

This is expected to leverage our existing understanding of rare disease and promises significant strides towards precision medicine and improving the lives of rare disease patients including those living with cystinosis.

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Editor Terri Schleuder

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 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.