cystinosis magazine

TOGETHER WE SHINE BRIGHT

A simple wish
– scribbled on the back of a napkin –
unequivocally became the wish of every
human affected by cystinosis.

The Wish Effect

Today,
Natalie Stack's wish has continued to
be the energetic drive for researchers,
friends, and supporters of the
Cystinosis Research Foundation
from around the world.

FOR FRIENDS AND SUPPORTERS OF THE CYSTINOSIS RESEARCH FOUNDATION

FALL 2021

The Wish Effect

We are grateful for everything we know. Everything we are. Everything we have. Everything we will be.

2003

- Natalie Stack made a wish on the eve of her 12th birthday, "to have my disease go away forever."
- The Cystinosis Research Foundation was established with the sole purpose of raising funds to find better treatments and a cure for cystinosis.

2013

- FDA approval in 2013 for a delayed-release form of cysteamine. CRF funded every early clinical study that led to the discovery of the delayed-release form of the medication now known as Procysbi®.
- First patient pilot study for an allogeneic stem cell study at UCLA.

2018

- FDA approval on December 19, 2018 for first stem cell and gene therapy clinical trial to test a new treatment for cystinosis.

2019

- First patient in stem cell and gene therapy clinical trial transplanted on October 7, 2019.

2020

- Second patient in stem cell and gene therapy clinical trial transplanted on June 29, 2020.
- Third patient in stem cell and gene therapy clinical trial transplanted on November 16, 2020.

2021

- Fourth patient in stem cell and gene therapy clinical trial transplanted on November 15, 2021.
- **Road to a cure!** Today, CRF is the largest fund provider of grants for cystinosis research in the world, issuing 207 grants in 12 countries.
- CRF has raised nearly $62 million, with 100% of your donations going to support cystinosis research. CRF's efforts have changed the course of cystinosis and given new energy to its investigators and scientists.
- CRF partnered with Sanford CoRDS to create the new Cure Cystinosis International Registry *(CCIR)*, the only international cystinosis patient registry in the world.
- CRF's commitment to research has given hope and promise to the global community of cystinosis patients and their families.

Dream. Wish. Purpose.

FALL 2021

THE WISH EFFECT

In 2003, Natalie Stack made a wish to find the cure for cystinosis. What started as a simple wish, has amassed into the wish of every human affected by cystinosis. This Wish Effect set a global community on the path to research success. Today, we celebrate CRF's legacy of community and research excellence. Together, We Shine Bright!

CONTACT US:
Please send suggestions and comments regarding *Cystinosis Magazine* to *nstack@cystinosisresearch.org*.

To receive our e-newsletter, *Star Facts*, send your email address to *zsolsby@cystinosisresearch.org*.

The entire cost of *Cystinosis Magazine* is underwritten by friends of the Cystinosis Research Foundation.

Art Direction and Printing: Idea Hall

cystinosis RESEARCH FOUNDATION

CYSTINOSISRESEARCH.ORG

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Irvine, California 92612

949 223 7610

The mission of the Cystinosis Research Foundation is to find better treatments and a cure for cystinosis by supporting bench, clinical and translational research. Since 2003, CRF has raised nearly $62 million for cystinosis research in an effort to find a cure.

cystinosis RESEARCH FOUNDATION

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CYSTINOSIS MAGAZINE IS A PUBLICATION OF THE CYSTINOSIS RESEARCH FOUNDATION

Dear Family & Friends

The year is coming to an end and for most of us, that is a welcome reality! Once again, it has been a year of challenges and setbacks but we have navigated the tumultuous waters and have persevered. The pandemic taught us how to be more creative, efficient and resilient. As a community, we have much to be grateful for and appreciate. The wind is at our backs and life is returning to normal – a new normal but one that brings a unique perspective on life and community.

THE WISH EFFECT

In 2003, on the eve of Natalie's 12th birthday, I asked her if she had a birthday wish. She responded with a wish that would change the course of our lives and propel us into a new world; a world filled with hope and promise. Natalie's wish, "to have my disease go away forever" was the start of our family's journey that now includes all of you. How fortunate we are to have all of you in our lives! The devastating diagnosis of cystinosis unexpectedly and gratefully brought us many blessings.

I often reflect on the day Natalie made her wish – a day seared into my memory. I can still remember reading the words she'd scribbled on the napkin and then holding her as I promised to make her wish come true. It didn't take long before we realized that Natalie's wish was the wish of all those with cystinosis and their families.

The wish, made by one little girl, had a ripple effect that altered the course of how we think about cystinosis. Natalie's wish positively impacted the global cystinosis community and inspired families to join us in our quest to find a cure for cystinosis. Her wish replaced the fear and uncertainty so many patients and families had, with hope. There is strength in numbers and together, we have accomplished what many thought was impossible – a new FDA approved treatment, an FDA approved clinical trial and the establishment of a global research community. Natalie's wish had an effect that resonated around the world.

RESEARCH IS OUR GUIDING LIGHT

Although the pandemic caused some delays in our research program, overall, the studies are back on track, moving forward and our researchers are making progress. We have built a collaborative, thriving international research community made up of the most brilliant, committed researchers who have dedicated their careers to our children and adults with cystinosis. They know our hope for a brighter future hinges on their work and discoveries.

One of the most significant consequences of CRF's commitment to support novel research ideas is that it has allowed new investigators to establish proof of concept. Many of those early studies have resulted in significant new discoveries allowing the researches to seek other funding sources thereby leveraging CRF's initial grant awards. Leveraging CRF's initial awards has accelerated the research process and increased our knowledge about cystinosis. Our strategic approach to research and our emphasis on collaboration has expanded the field of cystinosis and our understanding of the disease.

As a direct result of your unwavering support, CRF is the largest fund provider of cystinosis research in the world. Our research projects have led to new discoveries about cystinosis, new clinical trials and two FDA approvals. CRF has issued 207 multi-year grants, eight extension grants and awarded nine equipment grants. CRF-funded researchers have published 91 articles in prestigious research and medical journals. Our success is a direct result of your support and partnership with us to fund research that will lead to a cure.

UPDATE ON THE STEM CELL AND GENE THERAPY CLINICAL TRIAL

Since 2007, CRF has proudly supported and funded Stéphanie Cherqui, PhD, at UC San Diego for her groundbreaking stem cell and gene therapy research. At the time when Dr. Cherqui was seeking financial support for her research, there were very few resources available to her because her research was considered "speculative" and high risk. When CRF learned about her idea to use stem cells as a possible one-time treatment for cystinosis, we were intrigued and excited. CRF took the risk, partnered with her and has awarded over $5.78 million in grants for her research. As a result of our investment, Dr. Cherqui has received over $21.5 million in funding from other sources including CIRM and the NIH. And of course, the most impressive result has been the FDA approved of a clinical trial to test Dr. Cherqui's stem cell and gene therapy treatment. We are proud and honored to partner with Dr. Cherqui.

The stem cell trial is ongoing and we are happy to report that the fourth patient was transplanted on November 15, 2021. It was over two years ago that Jordan Janz became the first patient to volunteer for the stem cell study. On June 29, 2020, the second patient was transplanted and on November 16, 2020, Jacob Seachord became the third patient to be transplanted. We anticipate that by mid-year 2022, the final two patients will be transplanted and at that point, the trial will likely expand to include more patients in multiple locations. Please read more about the trial on page 16.

We are grateful to the four patient volunteers for blazing the path toward the cure. We are pleased to report that all three transplanted patients are doing extraordinarily well and have cystine levels low enough that they do not need to take cysteamine therapy. The genetically repaired cells are doing their job!

The reality is that very few research ideas make it from the bench to the bedside. The road to new treatments is long and arduous, fraught with setbacks, failures and disappointments. However, when you have a determined research community like we have, wishes and ideas become reality.

2021 RESEARCH GRANTS AWARDED - $612,243

In order to ensure that your donations are always at work, CRF awards new grants biannually. We are pleased to announce that in the spring of 2021, CRF awarded two research grants and one grant for lab equipment for a total amount of $612,243. The grants were awarded to researchers in Italy, Switzerland and the United States. Please read about the new grants awarded and learn more about the important research we are funding on page 27. The grant abstracts are listed on the following pages.

We have a fascinating article about Dr. Sophie Molholm, at the Albert Einstein College of Medicine who in collaboration with her colleagues, Drs. Ana Alves Francisco and John Foxe, are investigating how cystinosis affects brain function over time. Dr. Molhom's team will seek answers to how the disease impacts hearing, visual processing and memory, among other markers of brain function. Her exciting and understudied area of research will bring much-needed information about the brain and cystinosis. The study seeks to understand the possible cognitive effects of cystinosis, with the ultimate goal of improving the quality of life of children and adults with cystinosis. Please read more about Dr. Molholm's study on page 20.

THE EXPANSIVE IMPACT OF CRF RESEARCH

Cystinosis research has an impact on other more prevalent diseases and disorders. Discoveries made by CRF-funded researchers are being applied to Friedreich's Ataxia, Danon disease, corneal diseases, kidney diseases and genetic and systemic diseases similar to cystinosis. The success of our research program is a direct result of the leadership and commitment of our CRF Scientific Review Board who work diligently throughout the year reviewing the merits of every research application we receive and recommending only the best and most promising studies for funding. We thank the SRB (listed on page 31) for their guidance and dedication to the CRF research program and the cystinosis community.

OUR GRATITUDE RUNS DEEP

It is with an enormous amount of gratitude that we thank everyone who contributed and shared their story with us in this issue of the magazine. We have family stories from all over the world and each story is heartwarming and powerful. Sharing the cystinosis journey through the eyes of our families and patients helps us understand the uniqueness of each story and connects us in the most human, loving way.

We know that cystinosis impacts us every day and in turn, CRF is relentless in our pursuit of finding a cure every day. Our cystinosis community is part of the wish effect – we are more than hope alone, we are determined to make a difference in a meaningful and tangible way so that one day cystinosis will be a disease of the past. The research we fund will result in more breakthroughs, life-changing treatments, clinical trials and discoveries in the years to come.

We have faced the challenges of cystinosis together, we have overcome obstacles and together we have found a path to better treatments and a cure. Although we have made progress, our job is not done; there is more work to do and with your help and support, we will undoubtedly succeed. We are excited to resume our in-person Day of Hope family conference March 31 – April 2 and our spectacular annual gala, Natalie's Wish, on April 2 next year. We look forward to seeing you and celebrating our patient and research community.

Our deepest gratitude for your steadfast support of our research program and for your commitment to making Natalie's wish come true. You have been by our side while on this journey and you have lifted us up when we were challenged and forging new, sometimes uncertain, paths. It has been your encouragement and unwavering partnership that has allowed CRF to soar to heights that once were unimaginable.

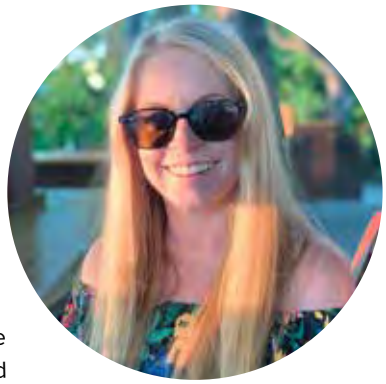
With heartfelt thanks and gratitude,

Nancy & Jeff

Dream. Wish. Purpose.

A Note from Natalie

Dear family and friends,



This year has been different, but getting better because the pandemic seems to be slowing down. I have been able to travel more, see more people, and am able to go into the office more frequently. It has been nice to start getting back to normal, but I know the world has changed in many ways because of the pandemic. Though I am fully vaccinated, it is scary to know that many people can still get the virus and get a severe case of it. I look forward to getting my booster shot soon so that I am even more protected. I also look forward to the day when we can all reunite and celebrate the success of the Cystinosis Research Foundation.

So far, 2021 has been a good year. In addition to turning 30 this year, I have continued to work at CASA (Court Appointed Special Advocates). I still enjoy living in Irvine, California with my chatty cat, Henry. Overall, my health remains stable and I have not had a kidney transplant yet although the older I get, the more likely it is that I will need a kidney transplant. I try to maintain my health by exercising regularly, eating healthy, sleeping at least eight hours each night, and taking my medication daily.

Cystinosis is a disease that constantly takes the energy out of you because of the round-the-clock medications that cause daily and extreme side effects from those medications. It is not easy living with cystinosis. The medications, severe fatigue, nausea and a myriad of other complications make it an exhausting disease and the older I get the more tired I feel. Though it does not look like it on the outside, my body is continuously and slowly shutting down. No matter how religious I am with my medications, the disease is always one step ahead of me.

I hope to participate in the stem cell clinical trail in the future. The data from the patients who have already participated shows a slowing in the severity and progression of the disease. I have hope that if I am transplanted, I will have a better quality of life and not have to worry as much about my future and deterioration of my health. The progress and success of the stem cell trial so far has allowed me believe that a brighter future lies ahead for all of us.

The cystinosis community has given me hope through all these years that I will one day be able to live a normal, healthy life. Words cannot express how thankful I am to the community and the doctors who never give up hope on finding a cure for cystinosis. Because of the work that the doctors, especially Dr. Cherqui, have done for cystinosis, I believe that my future is promising.

I am incredibly appreciative of our doctors, my parents and the rest of this community. Thank you for never giving up on my wish to have my disease go away forever.

Love, Natalie

cystinosis RESEARCH FOUNDATION | TOGETHER WE SHINE BRIGHT

What is cystinosis?

Cystinosis is a rare, inherited, metabolic disease that is characterized by the abnormal accumulation of the amino acid cystine in every cell in the body. Build-up of cystine in the cells eventually destroys all major organs of the body, including the kidneys, liver, eyes, muscles, bone marrow, thyroid and brain. Medication is available to control some of the symptoms of this terrible disease, but cystinosis remains incurable. Cystinosis affects approximately 600 people, mostly children, in North America, and about 2,000 worldwide.

It is one of the 7,000 rare or "orphan" diseases in the United States that collectively impact approximately 30 million Americans.

Federal funding for research on cystinosis and other rare diseases is virtually non-existent and most pharmaceutical companies remain uninterested because financial rewards are too small. Yet, while there are only a small number of patients who suffer from any given "orphan" disease, knowledge gained by studying one disease often leads to advancements in other rare diseases and more prevalent and well-known disorders.

Our story

In 2003, Natalie Stack made a wish on the eve of her 12th birthday, "to have my disease go away forever." That same year, the Cystinosis Research Foundation (CRF) was established with the sole purpose of raising funds to find better treatments and a cure for cystinosis.

Today, CRF is the largest fund provider of grants for cystinosis research in the world, issuing 207 grants in 12 countries.

CRF has raised nearly $62 million, with 100% of your donations going to support cystinosis research. CRF's efforts have changed the course of cystinosis and given new energy to its investigators and scientists. CRF's commitment to research has given hope and promise to the global community of cystinosis patients and their families.

CHARITY NAVIGATOR ★★★★ Four Star Charity

GUIDESTAR PLATINUM TRANSPARENCY 2021

We celebrate our CRF community and are grateful every day for your support.

CRF's highly strategic approach to funding has resulted in two FDA approvals and several human clinical trials. The research dollars we have invested have been leveraged by over $28 million in grants from other funding agencies. Not only does CRF research help our community, but our discoveries are applied to more prevalent diseases and disorders. CRF-funded research has the potential to help millions of others.

We want to thank our families, friends and donors who have remained steadfast in their commitment to finding better treatments and a cure. Thank you to the cystinosis researchers and scientists who are working around the clock on behalf of our children and adults with cystinosis.

Thank You!

You have changed the course of cystinosis!

In 2021

3 New Studies Funded

2 New Research Grants

1 Equipment Grant

Totaling More Than **$612,000**

CRF partnered with Sanford CoRDS (Coordination of Rare Diseases at Sanford) **to create the new Cure Cystinosis International Registry** (CCIR), **the only international cystinosis patient registry in the world.**

100%

Of Your Donations Directly Support Cystinosis Research

1 FDA Approved Drug

1 FDA Approved Clinical Trial

Since 2003

207 Multi-Year Grants Funded In **12** Countries

92 Articles Published In Prestigious Journals By CRF Researchers

Nearly **$62MM** Raised For Cystinosis Research

Save The Date

CYSTINOSIS CURESEARCH FOUNDATION

Together, We Shine Bright!

Learn, share, laugh and celebrate our cystinosis community at the Day of Hope family conference. We will renew our friendships and our efforts to work together to support research that will improve the lives of our children and adults with cystinosis. CRF researchers will present their current research findings and report on their progress. We will have updates on the stem cell trial, and we will discuss current research focused on muscle wasting, cystinosis and brain function, and a potential novel treatment for corneal cystinosis.

After two long years apart, we eagerly await seeing you and renewing and strengthening the bonds that created this remarkable CRF family community.
More information coming soon!
Save the date – see you in 2022!

WHEN

Thursday, March 31, 2022 –
Saturday, April 2, 2022

WHERE

Balboa Bay Resort
Newport Beach, CA

For more information contact info@cystinosisresearch.org / 949 223 7610

Save The Date

Natalie's 2022 Wish Celebration

together again

we've dreamed
we've wished
we've changed

let's share
let's celebrate
together again

Natalie's Wish Celebration

Saturday, April 2, 2022

Balboa Bay Resort, Newport Beach, CA

Please join us in honoring the children and adults who are affected by cystinosis and the cystinosis research community for its commitment to our children. We look forward seeing you on Saturday, April 2, 2022!

For more information contact Zoe Solsby *zsolsby@cystinosisresearch.org* / 949 223 7610

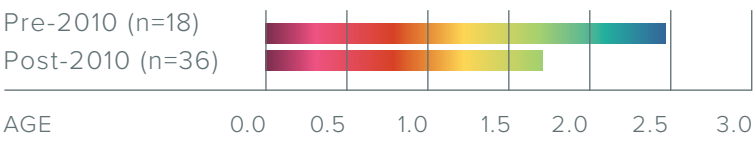
The new **Cure Cystinosis International Registry** (CCIR) was launched in the Spring of 2021.

Introduction

Considering the ultra-rare nature of the disease, with only an estimated 600 patients in the United States, individual input from patients is extremely important and useful. Furthermore, individuals with cystinosis often experience widely different symptoms and complications requiring unique and individualized treatment and care. This further illustrates the significance of participation in the registry. The registry is a key link between patients and researchers, which ultimately both enables and guides research on improved treatment for cystinosis. Your participation in the patient registry can help accelerate research towards improved treatment and a cure.

Since the launch of the new registry, patients from 14 countries have participated in the registry. Due in part to the extremely low incidence of the disease and the myriad of initial symptoms, preliminary data from participants has shown that cystinosis continues to be a difficult disease to diagnose. Initial diagnosis for patients without an affected family member have only slightly decreased in recent decades, with an average age of diagnosis in years prior to 2010 being 2.5 years compared to 1.7 years since 2010.

AVERAGE AGE OF DIAGNOSIS



By Clay Emerson, PhD
CCIR Committee Member
and Brooke Emerson's dad
Hammonton, New Jersey, USA

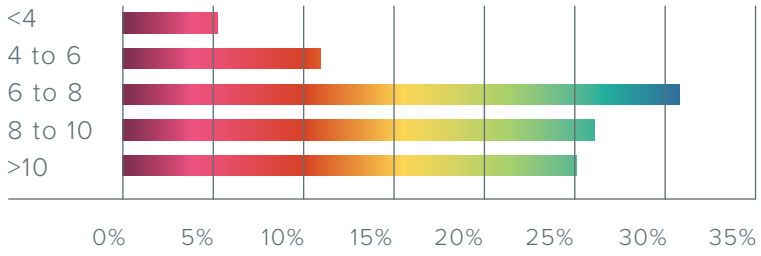
> > >

Medications

People with cystinosis often must take numerous medications in order to manage the many manifestations of cystinosis. Most of these medications require adherence to strict dosage timing and some must be taken in a manner such that they do not interfere with other necessary medications. This results in a complex medication schedule. Current data from the CCIR indicates that most patients take more than eight different types of medication throughout the day with some participants taking as many as 13 different types of medication requiring up to 25 individual doses per day.

Cure Cystinosis
International Registry

NUMBER OF MEDICATIONS

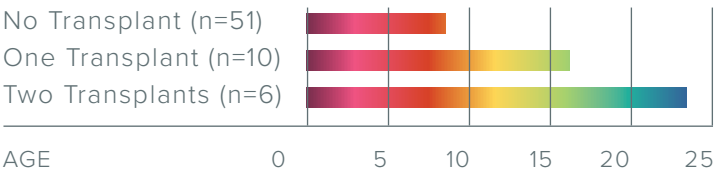


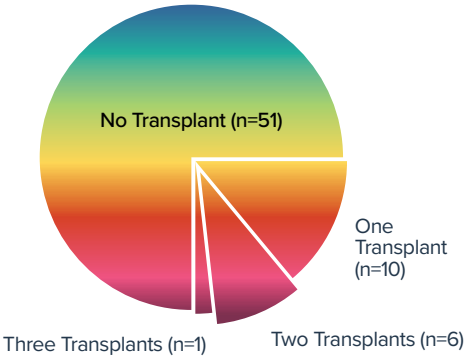
Kidney Transplant

Kidneys are typically the first organ which are impacted by cystinosis, and kidney transplant is a routine intervention for people with cystinosis. The donation of a kidney can be life changing for patients with cystinosis. If you are interested in being an organ donor, please consider registering at *www.organdonor.gov.* The new patient registry has further confirmed how important kidney transplant is for people with cystinosis. Every patient's journey with cystinosis is unique, and the need for a kidney transplant arises at very different times from one person to another. The need for multiple transplants is not uncommon, with 10% of current participants in the registry having had multiple (two or three) transplants to date. Early participation in the CCIR indicates that approximately 75% of respondents have not yet had a kidney transplant. The average age of participants who have not yet had a transplant was 9 years old (n=51). The average age for a first transplant was 16 years and the average age for the second transplant was 24 years.

EVERY PATIENT'S JOURNEY WITH CYSTINOSIS IS UNIQUE

AVERAGE AGE OF KIDNEY TRANSPLANT





CCIR CONTINUED >>>

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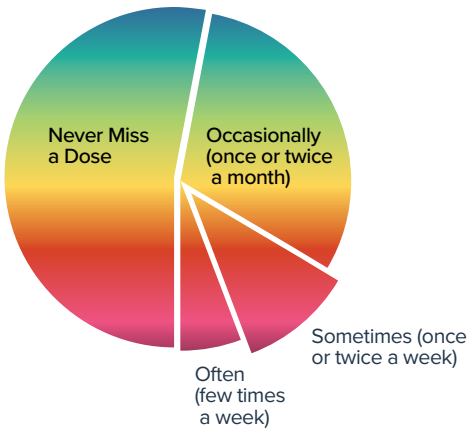
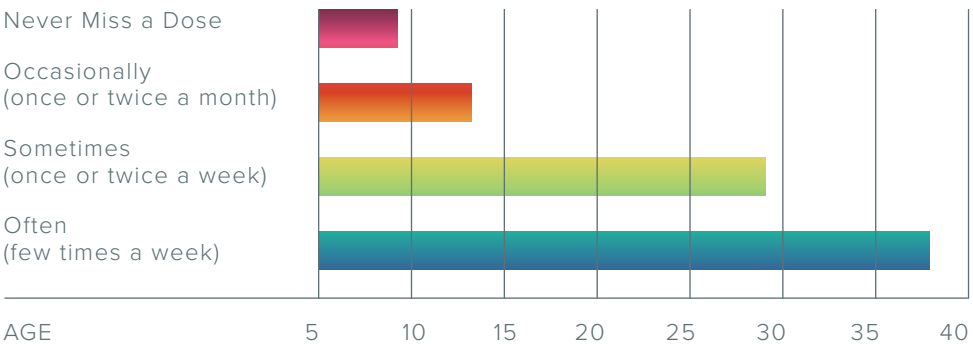
The new **Cure Cystinosis International Registry** (CCIR) was launched in the Spring of 2021.

Cysteamine Compliance

The availability of the drug cysteamine changed the natural course of the disease and currently is the only approved treatment for cystinosis. Unfortunately, therapeutic treatment with cysteamine requires a relatively high dose and must be administered around-the-clock at either a 6-hour (Cystagon®) or 12-hour (Procysbi®) dosing frequency. Cysteamine also has well-documented side effects which include but are not limited to nausea, loss of appetite, abdominal pain and both breath and body odor. Cysteamine has been shown to delay end-stage renal failure and the need for a kidney transplant. Despite currently being the only way to manage the progression of the disease, the combination of a strict dosing schedule (often including a nighttime administration) and extremely common and sometimes severe side effects, make routine and strict compliance with cysteamine treatment very difficult for many patients. Early information from the CCIR shows that nearly half of participants (47%) reporting some level of difficulty in compliance with cysteamine therapy. Compliance is often especially difficult as children transition from pediatric care to more independent adult care. Similarly, social, school, and career pressures often make compliance even more difficult. Current data from the new CCIR illustrate this reality and the need for better treatment. The average age of respondents reporting good and strict compliance was only 9 years, while the average age of those reporting that they sometimes (once or twice a week) have difficulty with compliance being 29 years.



CYSTEAMINE NON-COMPLIANCE WITH AGE



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Conclusion

In less than its first year the new patient registry is already helping to inform researchers and ultimately accelerate the development of better treatments and a cure for cystinosis. The value of the registry will only truly be realized with patient participation. We strongly encourage patients or caregivers to participate in the registry and help identify the needs of patients with cystinosis and accelerate research. The questionnaire takes about 40 minutes to complete and registration is simple. Please visit the CRF website to sign up today!

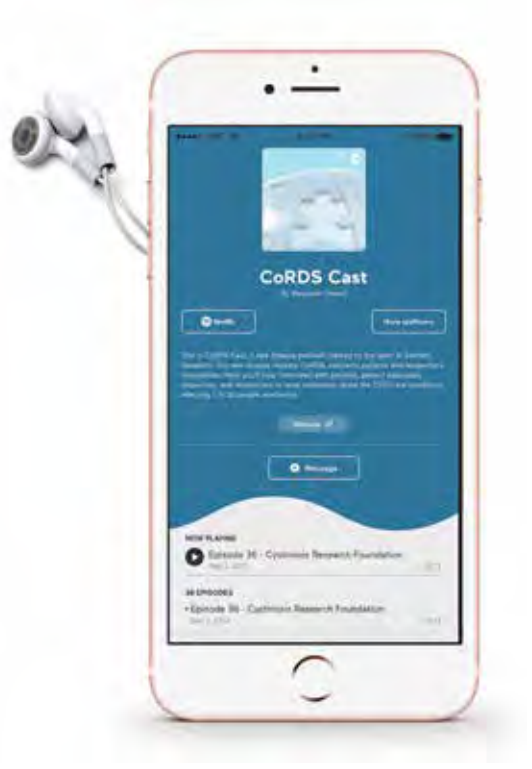
THE VALUE OF THE REGISTRY WILL ONLY TRULY BE REALIZED WITH PATIENT PARTICIPATION

www.cystinosisresearch.org/cure-cystinosis-international-registry

Listen to the Rare Disease Podcast Interview with Cystinosis Parents, Clay Emerson and Stephen Jenkins

Tune in to the latest episode of CoRDS Cast, a rare disease podcast created by Sanford Research, featuring Clay Emerson and Stephen Jenkins, their experiences as fathers of children with cystinosis, and their thoughts on the importance of joining the Cure Cystinosis International Registry. CRF partnered with CoRDS to create the Cure Cystinosis International Registry. When you enroll in CCIR and answer the patient questionnaire, your collective data will be an invaluable resource for the research community to learn more about cystinosis and its symptoms to develop better treatments and a cure.

FIND EPISODE 36 AT **https://anchor.fm/cords-cast**



CRF'S LEGACY OF
RESEARCH SUCCESS

RESEARCHERS
Stéphanie Cherqui, PhD AND
Corinne Antignac, MD, PhD
IN THE LAB

Research

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The Wish Effect

PDATE

ON THE CLINICAL TRIAL TO TEST STEM CELL AND GENE THERAPY APPROACH FOR CYSTINOSIS

AFTER OVER A DECADE OF RESEARCH, Stéphanie Cherqui, PhD, Associate Professor of Pediatrics at the University of California San Diego, launched the first-in-human stem cell gene therapy clinical trial for cystinosis. In cystinosis, the body is not able to properly clear the amino acid cystine leading to accumulation of cystine and cystine crystals in blood and tissues.

The new stem cell gene therapy approach pioneered by Dr. Cherqui for treating cystinosis involves infusion of a patient's own genetically modified stem cells. In this procedure, hematopoietic stem cells (blood cell precursors) are removed from a patient and engineered in the lab using gene therapy techniques to incorporate a functional copy of the CTNS gene, which is responsible for synthesis of the missing protein, cystinosin. Through a process called mobilization and apheresis, the patient's hematopoietic cells are obtained from the blood for genetic modification in the lab. After that, a lentiviral vector, which is a safe version of the HIV, is used to introduce the CTNS gene in the patient's stem cells. Then, the genetically modified cells are infused back to the same patient after conditioning which involves administering to the patient a chemotherapy agent designed to "make space" so that the genetically modified cells can engraft in the patient's bone marrow.

Once the modified cells engraft, they will potentially divide and differentiate, providing a population of corrected cells with the normal CTNS gene. Those cells are then expected to circulate in the blood and migrate throughout the patient's body to damaged tissues and organs, where they will produce the normal cystinosin protein and potentially begin reducing cystine levels.

The Phase 1/2 clinical trial (clinicaltrials.gov NCT03897361) evaluates the safety and efficacy

of this stem cell gene therapy approach. Patients are assessed for eligibility for the trial participation, including evaluation of organ function and cystine levels in blood and various tissues. Oral cysteamine and cysteamine eye drops are withdrawn prior to stem cell infusion.

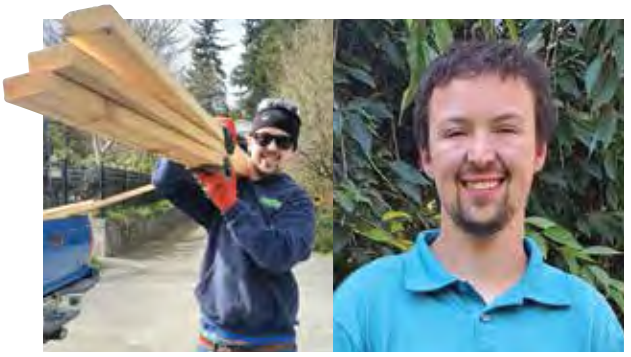
Three patients (ages 20 to 46 years) have been treated with this approach so far. Their follow-up appointments range from 6 months to 18 months. There have been no adverse events related to the drug product, and no serious adverse events have been reported to date. Successful bone marrow recovery was obtained in all three patients. The genetically modified cells were present in the peripheral blood of all three patients. White blood cell cystine was decreased in all three treated patients. In addition, results show decrease in tissue cystine crystals in the skin, eyes, and rectal mucosa in Patient #1 at 18-months post gene therapy.

All three patients are no longer taking oral cysteamine. Patient #2 has restarted cysteamine eyedrops as some buildup of crystals in the cornea was noted at the 12-month post-gene therapy appointment; this is being carefully monitored. Patients will be followed in a long-term follow-up study to further evaluate the therapy's safety and potential efficacy.

I am currently enrolled in the Butchery and Charcuterie program at Southern Alberta Institute of Technology (SAIT) in Calgary. I have been in school since September 1st and I am loving it! I'm making lots of new friends and learning lots of new things. I am hoping that I will start my new career once I am done with the program at SAIT in April 2022. I plan to own my own business one day.

I have been feeling great lately and I think a fresh start in a different city has really helped my mindset. I feel amazing; I am getting my energy back at last, and my cystine levels are still promising.

- JORDAN JANZ



Since the time of the stem cell transplant, I have been feeling great and I feel like I have more energy now. One of the best things about the transplant is that I do not have to take as much medicine as before.

I have been working for my parent's construction company, but I am also looking for a job as an assistant physical therapist. My family and I have been having fun going camping with our friends. The transplant was hard to go through but I believe it was all worth it.

- JACOB SEACHORD

FINAL STUDY REPORT

Olivier Devuyst, MD, PhD
University of Zurich
Zurich, Switzerland

Francesco Emma, MD
Bambino Gesù Children's Hospital
Rome, Italy

IN 2018 CRF awarded a grant to **Olivier Devuyst, MD, PhD**, and **Francesco Emma, MD**, for their research on the **DEVELOPMENT AND CHARACTERIZATION OF A RAT MODEL OF CYSTINOSIS.** They successfully generated a novel *CTNS-/-* rat model that replicates the clinical symptoms of human nephropathic cystinosis. The *CTNS-/-* rat model will be an invaluable tool for the research community.

DOES CYSTINOSIS AFFECT BRAIN FUNCTION?

NEW STUDY SEEKS TO PROVIDE ANSWERS

THANKS TO CRF SUPPORT, NEUROSCIENTIST SOPHIE MOLHOLM, PhD, AND HER COLLEAGUES AT ALBERT EINSTEIN COLLEGE OF MEDICINE WILL EXPLORE POSSIBLE LONG-TERM IMPACTS ON MEMORY AND FOCUS.

By Dennis Arp

Brain researcher, **DR. SOPHIE MOLHOLM**, has no shortage of patient populations to study as she explores developmental disorders and how they impact the quality of life. There are clinical reasons why she chose cystinosis for her new study, but science isn't her only source of motivation.

Receiving support from the Cystinosis Research Foundation (CRF) and working with cystinosis families and physicians has her excited about the progress the community can make together.

"Getting involved with a foundation like the CRF, we really have a chance to become part of this community," said Dr. Molholm, professor in the Departments of Pediatrics, Neuroscience, and Psychiatry & Behavioral Sciences at Albert Einstein College of Medicine in New York City.

"It's inspiring to see something build and become so sophisticated, making a huge difference in the lives of individuals," added Dr. Molholm, who is also co-director of Einstein College's Rose F. Kennedy Intellectual & Developmental Disabilities Research Center and the Cognitive Neurophysiology Laboratory.

A grant from the CRF makes it possible for Dr. Molholm, in collaboration with her colleagues Drs. Ana Alves Francisco and John Foxe, to investigate how cystinosis affects brain function over time. By performing tests with patients in their New York offices and lab, the team will seek answers to how the disease impacts hearing, visual processing and memory, among other markers of brain function.

Previous studies suggest a possible link between cystinosis and loss of short-term memory. That's one reason this is a good time for a study that takes a more focused look at those connections so treatment plans can be developed and refined, Dr. Molhom said.

"This work will reveal novel information on how brain function is affected by cystinosis," she explained. "It will add to what we currently know by testing if what we consider to be part of the profile of cystinosis is mainly associated with the mutation, or if it is more generally seen in people with kidney dysfunction. We will do thorough characterizations of cognitive and neural function, identify strengths and weaknesses, and determine brain markers that can be used to assess the effectiveness of treatments."

Dr. Molholm brings more than 15 years of experience to her research work, which has focused largely on understanding the neurobiology of developmental disorders, with an emphasis on autism. Her lab's tools include non-invasive high-density recordings of electrical activity in the brain, as well as magnetic resonance imaging (MRI).

"From a global research perspective, there isn't that much information on how brain function is affected by cystinosis," the researcher said. "So we can use these tools to very precisely measure neurocognitive function."

Providing details about the possible cognitive effects of cystinosis can have a profound effect on the lives of patients and their real-world experiences, Dr. Molholm said.

"You might say, 'What does it matter to have small problems with your sensory memory?'" she said. "But it's these things that feed into more complex brain processes. So if something is not working at these fundamental stages, it means patients have to work harder at later stages. The impact is felt in their ability to do schoolwork, to stay focused in the classroom or at their jobs. Adults have similar concerns – it's not just children."

The study is divided into two parts, explained Co-Investigator Dr. Ana Alves Francisco, research assistant professor at the Albert Einstein College of Medicine and the person who is in direct contact with participating families.

"There's the EEG part, in which the participant performs tasks like clicking a button when they see an image while we record their brain electrical activity," Dr. Alves Francisco said.

"Then there's also neuropsychological testing when we're able to speak with participants and characterize thoroughly weaknesses and strengths of cognitive function – verbal and nonverbal abilities, memory and such."

To deliver the best results possible, the study team is enrolling 25 cystinosis patients, 25 unaffected control participants, and 25 patients with chronic kidney disease. Patients of all ages are eligible, and enrollment is ongoing, Dr. Alves Francisco said.

The team has started working with some study participants as it continues to recruit, recognizing the complications related to the COVID-19 pandemic will lengthen the time of the study. She and her teammates are taking extra steps to ensure that families have a positive experience during their two- to three-day stays in New York for testing and some fun activities in the city, when possible.

In the end, the researchers will publish the findings from these studies in the peer-reviewed scientific literature, so that families, clinicians, and other researchers can benefit from the knowledge gained under this project.

Dr. Alves Francisco has been working with cystinosis patients and families for about five years now, and she says one reason she's so excited about this research project is that she has found that even young cystinosis patients "are motivated to participate because they want the community and those who come after them to enjoy a better life."

"These kids have so many complications in their lives, and yet they're willing to do more than is asked of them so they can help others," she added. "That's something beautiful I've encountered in the cystinosis community, and it motivates me to do this work."

Dr. Molholm adds that the work wouldn't happen without the support of the Cystinosis Research Foundation.

"CRF funding is tremendously meaningful for us, as it's also meaningful to be involved with such committed people who bring this community together," she said. "Seeing all the love and energy that goes into it and the amazing amount of knowledge that results, all of these factors provide us with great inspiration."

THE IMPACT OF CRF RESEARCH

AREAS OF RESEARCH FOCUS & GRANTS AWARDED SINCE 2002

New Drug Discovery Cysteamine, New Medications and Devices

30 GRANTS

Ghanashyam Acharya, PhD
BAYLOR COLLEGE OF MEDICINE, HOUSTON, TEXAS

Francesco Bellomo, PhD
Francesco Emma, MD
BAMBINO GESÙ CHILDREN'S HOSPITAL, ROME, ITALY

Pierre Courtoy, MD, PhD
Christophe Pierreux, PhD
DE DUVE INSTITUTE, LOUVAIN UNIVERSITY MEDICAL SCHOOL, BRUSSELS, BELGIUM

Laura Rita Rega, PhD
BAMBINO GESÙ CHILDREN'S HOSPITAL, ROME, ITALY

Antonella De Matteis, MD
TELETHON INSTITUTE OF GENETICS AND MEDICINE, NAPLES, ITALY

Ranjan Dohil, MD
UNIVERSITY OF CALIFORNIA, SAN DIEGO, LA JOLLA, CALIFORNIA

Francesco Emma, MD
Laura Rita Rega, PhD
BAMBINO GESÙ CHILDREN'S HOSPITAL, ROME, ITALY

Paul Goodyer, MD
MONTRÉAL CHILDREN'S HOSPITAL, MONTRÉAL, QUÉBEC, CANADA

Jennifer Hollywood, PhD
Alan Davidson, PhD
UNIVERSITY OF AUCKLAND, AUCKLAND, NEW ZEALAND

Michael Sekar, PhD
AMMA THERAPEUTICS, INC., HAYWARD, CALIFORNIA

Laura Rita Rega, PhD
BAMBINO GESÙ CHILDREN'S HOSPITAL, ROME, ITALY

Vincent Stanton, Jr., MD
Patrice Rioux, MD, PhD
THIOGENESIS THERAPEUTICS, INC., SAN DIEGO, CALIFORNIA

Eye-Corneal Cystinosis Research

10 GRANTS

Ghanashyam Acharya, PhD
BAYLOR COLLEGE OF MEDICINE, HOUSTON, TEXAS

Stéphanie Cherqui, PhD
UNIVERSITY OF CALIFORNIA, SAN DIEGO, LA JOLLA, CALIFORNIA

Morgan Fedorchak, PhD
Kanwal Nischal, MD, FRCO
UNIVERSITY OF PITTSBURGH SCHOOL OF MEDICINE, PITTSBURGH, PENNSYLVANIA

Jennifer Simpson, MD
UNIVERSITY OF CALIFORNIA, IRVINE, IRVINE, CALIFORNIA

Kang Zhang, MD, PhD
UNIVERSITY OF CALIFORNIA, SAN DIEGO, LA JOLLA, CALIFORNIA

Cystine Measurement and Cysteamine Toxicity Study

10 GRANTS

Bruce Barshop, MD, PhD
UNIVERSITY OF CALIFORNIA, SAN DIEGO, LA JOLLA, CALIFORNIA

Shawn Davidson, PhD
PRINCETON UNIVERSITY, PRINCETON, NEW JERSEY

Thomas Jeitner, PhD
NEW YORK MEDICAL COLLEGE, VALHALLA, NEW YORK

Elena Levtchenko, MD, PhD
UNIVERSITY HOSPITAL, LEUVEN, BELGIUM

Cellular and/or Molecular Studies of the Pathogenesis of Cystinosis

59 GRANTS

Corinne Antignac, MD, PhD
IMAGINE INSTITUTE (INSERM U1163), PARIS, FRANCE

Francesco Bellomo, PhD NEW
Francesco Emma, MD
BAMBINO GESÙ CHILDREN'S HOSPITAL, ROME, ITALY

Sergio Catz, PhD
Raquel Carvalho Gontijo, PhD
THE SCRIPPS RESEARCH INSTITUTE, LA JOLLA, CALIFORNIA

Sergio Catz, PhD
Farhana Rahman, PhD
THE SCRIPPS RESEARCH INSTITUTE, LA JOLLA, CALIFORNIA

Sergio Catz, PhD
Nadia Zgajnar, PhD
THE SCRIPPS RESEARCH INSTITUTE, LA JOLLA, CALIFORNIA

Antonella De Matteis, MD
TELETHON INSTITUTE OF GENETICS AND MEDICINE, NAPLES, ITALY

Olivier Devuyst, MD, PhD
Zhiyong Chen, PhD
UNIVERSITY OF ZÜRICH, ZÜRICH, SWITZERLAND

Olivier Devuyst, MD, PhD
Alessandro Luciani, PhD
UNIVERSITY OF ZÜRICH, ZÜRICH, SWITZERLAND

Liang Feng, PhD
Xue Guo, PhD
STANFORD UNIVERSITY, PALO ALTO, CALIFORNIA

Bruno Gasnier, PhD
Yann Terras, MSc
PARIS DESCARTES UNIVERSITY, PARIS, FRANCE

Taosheng Huang, MD, PhD
UNIVERSITY OF CALIFORNIA, IRVINE, IRVINE, CALIFORNIA

Elena Levtchenko, MD, PhD
UNIVERSITY HOSPITAL, LEUVEN, BELGIUM

Ming Li, PhD
Jacob Kitzman, PhD
UNIVERSITY OF MICHIGAN, ANN ARBOR, MICHIGAN

Alessandro Luciani, PhD
UNIVERSITY OF ZÜRICH, ZÜRICH, SWITZERLAND

Gennaro Napolitano, PhD
THE SCRIPPS RESEARCH INSTITUTE, LA JOLLA, CALIFORNIA

Norbert Perrimon, PhD
HARVARD MEDICAL SCHOOL, BOSTON, MASSACHUSETTS

Giusi Prencipe, PhD
BAMBINO GESÙ CHILDREN'S HOSPITAL, ROME, ITALY

Matias Simons, MD
Marelja Zvonimir, PhD
IMAGINE INSTITUTE, PARIS, FRANCE

Jess Thoene, MD
TULANE UNIVERSITY SCHOOL OF MEDICINE, NEW ORLEANS, LOUISIANA

Bruno Vogt, MD NEW
Daniel Pouly, PhD
UNIVERSITY HOSPITAL OF BERN, BERN, SWITZERLAND

Skin, Muscle and Bone

16 GRANTS

Justine Bacchetta, MD, PhD
Irma Machuca-Gayet, PhD
HOSPICES CIVILS DE LYON UNIVERSITÉ DE LYON, LYON, FRANCE

Robert Ballotti, PhD
Christine Chiaverini, MD, PhD
FACULTÉ DE MÉDECINE, NICE, FRANCE

Andrea Del Fattore, PhD
Giulia Battafarano, PhD
BAMBINO GESÙ CHILDREN'S HOSPITAL, ROME, ITALY

Paul Grimm, MD
STANFORD UNIVERSITY SCHOOL OF MEDICINE, PALO ALTO, CALIFORNIA

Mary Leonard, MD, MSCE
STANFORD UNIVERSITY SCHOOL OF MEDICINE, PALO ALTO, CALIFORNIA

Robert Mak, MD, PhD
UNIVERSITY OF CALIFORNIA, SAN DIEGO, LA JOLLA, CALIFORNIA

Richard Reimer, MD
Jacinda Sampson, MD, PhD
Mary Leonard, MD, MSCE
Paul Grimm, MD
Trinh Tina Duong, MPT
Feliks Kogan, PhD
STANFORD UNIVERSITY, PALO ALTO, CALIFORNIA

Reza Seyedsadjadi, MD
Florian Eichler, MD
Lee Rubin, PhD
MASSACHUSETTS GENERAL HOSPITAL, BOSTON, MASSACHUSETTS

Thyroid

1 GRANT

Pierre Courtoy, MD, PhD
DE DUVE INSTITUTE, LOUVAIN UNIVERSITY MEDICAL SCHOOL, BRUSSELS, BELGIUM

CONTINUED ON NEXT PAGE

CONTINUED FROM PAGE 23

AREAS OF RESEARCH FOCUS & GRANTS AWARDED SINCE 2002

Molecular Study of Cystinosis in the Yeast Model

3 GRANTS

Bruno André, PhD
UNIVERSITÉ LIBRE DE BRUXELLES, GOSSELIES, BELGIUM

Anand Bachhawat, PhD
IISER MOHALI, MANAULI, PUNJAB, INDIA

David Pearce, PhD
UNIVERSITY OF ROCHESTER MEDICAL CENTER, ROCHESTER, NEW YORK

Genetic Analysis of Cystinosis

5 GRANTS

Katy Freed, PhD
TEXAS BIOMEDICAL RESEARCH INSTITUTE, SAN ANTONIO, TEXAS

Sihoun Hahn, MD, PhD
SEATTLE CHILDREN'S HOSPITAL, SEATTLE, WASHINGTON

Elena Levtchenko, MD, PhD
UNIVERSITY HOSPITAL, LEUVEN, BELGIUM

Eric Moses, PhD
TEXAS BIOMEDICAL RESEARCH INSTITUTE, SAN ANTONIO, TEXAS

Minnie Sarwal, MD, PhD
UNIVERSITY OF CALIFORNIA, SAN FRANCISCO, SAN FRANCISCO, CALIFORNIA

Neurological

16 GRANTS

Angela Ballantyne, PhD
UNIVERSITY OF CALIFORNIA, SAN DIEGO, LA JOLLA, CALIFORNIA

Miriam Britt Sach, MD, PhD
UNIVERSITY OF CALIFORNIA, SAN DIEGO, LA JOLLA, CALIFORNIA

Rita Ceponiene, MD, PhD
UNIVERSITY OF CALIFORNIA, SAN DIEGO, LA JOLLA, CALIFORNIA

Florian Eichler, MD
MASSACHUSETTS GENERAL HOSPITAL, BOSTON, MASSACHUSETTS

Sophie Molholm, PhD

John Foxe, PhD
ALBERT EINSTEIN COLLEGE OF MEDICINE, BRONX, NEW YORK

Aude Servais, MD, PhD
NECKER HOSPITAL, PARIS, FRANCE

Amy Spilkin, PhD
UNIVERSITY OF CALIFORNIA, SAN DIEGO, LA JOLLA, CALIFORNIA

Doris Trauner, MD
UNIVERSITY OF CALIFORNIA, SAN DIEGO, LA JOLLA, CALIFORNIA

Cure Cystinosis International Registry (CCIR)

1 GRANT

Ranjan Dohil, MD
UNIVERSITY OF CALIFORNIA, SAN DIEGO, LA JOLLA, CALIFORNIA

Rat Model for Cystinosis

3 GRANTS

Francesco Emma, MD
BAMBINO GESÙ CHILDREN'S HOSPITAL, ROME, ITALY

Olivier Devuyst, MD, PhD
UNIVERSITY OF ZÜRICH, ZÜRICH, SWITZERLAND

THE IMPACT OF CRF RESEARCH

Stem Cells and Gene Therapy: Bone Marrow Stem Cells, Induced Pluripotent Stem Cells, Gene Therapy and Gene Editing

33 GRANTS

Stéphanie Cherqui, PhD
UNIVERSITY OF CALIFORNIA, SAN DIEGO,
LA JOLLA, CALIFORNIA

Alan Davidson, PhD
THE UNIVERSITY OF AUCKLAND,
GRAFTON,
AUCKLAND, NEW ZEALAND

Bruno Gasnier, PhD
PARIS DESCARTES UNIVERSITY,
PARIS, FRANCE

Paul Goodyer, MD
MONTRÉAL CHILDREN'S HOSPITAL,
MONTRÉAL, QUEBEC, CANADA

Patrick Harrison, PhD
UNIVERSITY COLLEGE CORK,
CORK, IRELAND

Vasiliki Kalatzis, PhD
INSTITUTE OF MOLECULAR
GENETICS OF MONTPELLIER,
MONTPELLIER, FRANCE

Winston Kao, PhD
Hassane Amlal, PhD
UNIVERSITY OF CINCINNATI,
CINCINNATI, OHIO

Daniel Salomon, MD
THE SCRIPPS RESEARCH INSTITUTE,
LA JOLLA, CALIFORNIA

Holger Willenbring, MD
UNIVERSITY OF CALIFORNIA,
SAN FRANCISCO,
SAN FRANCISCO, CALIFORNIA

Kidney Research

22 GRANTS

Robert Chevalier, MD
UNIVERSITY OF VIRGINIA,
CHARLOTTESVILLE, VIRGINIA

Pierre Courtoy, MD, PhD
Christophe Pierreux, PhD
DE DUVE INSTITUTE, LOUVAIN
UNIVERSITY MEDICAL SCHOOL,
BRUSSELS, BELGIUM

Olivier Devuyst, MD, PhD
UNIVERSITY OF ZÜRICH,
INSTITUTE OF PHYSIOLOGY,
ZÜRICH, SWITZERLAND

Allison Eddy, MD
BC CHILDREN'S HOSPITAL,
VANCOUVER, CANADA

Benjamin Freedman, PhD
UNIVERSITY OF WASHINGTON,
SEATTLE, WASHINGTON

Elena Levtchenko, MD, PhD
UNIVERSITY HOSPITAL,
LEUVEN, BELGIUM

Robert Mak, MD, PhD
UNIVERSITY OF CALIFORNIA,
SAN DIEGO,
LA JOLLA, CALIFORNIA

Tara McMorrow, MD
UNIVERSITY COLLEGE DUBLIN,
BELFIELD, DUBLIN, IRELAND

Philip Newsholme, PhD
CURTIN UNIVERSITY,
PERTH, WESTERN AUSTRALIA

Daryl Okamura, MD
SEATTLE CHILDREN'S
RESEARCH INSTITUTE,
SEATTLE, WASHINGTON

Laura Rita Rega, PhD
BAMBINO GESÙ CHILDREN'S
HOSPITAL, ROME, ITALY

Mary Taub, PhD
UNIVERSITY AT BUFFALO,
THE STATE UNIVERSITY OF NEW YORK,
BUFFALO, NEW YORK

Lab Equipment for Cystinosis

9 GRANTS

Ghanashyam Acharya, PhD
BAYLOR COLLEGE OF MEDICINE,
HOUSTON, TEXAS

Corinne Antignac, MD, PhD
IMAGINE INSTITUTE, PARIS, FRANCE

Bruce Barshop, MD, PhD
UNIVERSITY OF CALIFORNIA,
SAN DIEGO, LA JOLLA, CALIFORNIA

NEW

Sergio Catz, PhD
THE SCRIPPS RESEARCH INSTITUTE,
LA JOLLA, CALIFORNIA

Stéphanie Cherqui, PhD
UNIVERSITY OF CALIFORNIA,
SAN DIEGO, LA JOLLA, CALIFORNIA

FALL 2021

PUBLISHED STUDIES

CRF-FUNDED RESEARCHERS have been instrumental in advancing the field of cystinosis through the publication of articles in prestigious journals. Published articles enable other scientists, pharmaceutical companies and the cystinosis community to learn more about the pathogenesis of cystinosis, to explore ideas for novel treatments and to prepare for clinical trials. We congratulate all of the published CRF-funded researchers who have dedicated their careers to the children and adults with cystinosis.

Bruno Gasnier, PhD
Paris-Descartes University
Paris, France

Congratulations to Bruno Gasnier, PhD, whose article, **"Arginine-Selective Modulation of the Lysosomal Transporter PQLC2 through a Gate-Tuning Mechanism"** was recently published in the August 2021 issue of the *PNAS Journal (Proceedings of the National Academy of Sciences of the United States)*.

CRF has provided over $1 million in funding to Dr. Gasnier since 2007. Read the published study of his work to understand the relevance and treatment of cystinosis:

www.cystinosisresearch.org/wp-content/uploads/2021/08/Dr.-Gasnier-Arginine-selective-modulation-of-the-lysosomal-PNAS-Aug-10-2021.pdf

Robert Mak, MD, PhD
Rady Children's Hospital
University of California, San Diego

We are pleased to announce that Robert Mak, MD, PhD, Professor and Chief, Division of Pediatric Nephrology, Rady Children's Hospital, University of California, San Diego, has two recently published studies.

"Targeting Interleukin-1 for Reversing Fat Browning and Muscle Wasting in Infantile Nephropathic Cystinosis," appeared June 2021 in the *Journal of Cachexia, Sarcopenia and Muscle*, and **"A Leptin Receptor Antagonist Attenuates Adipose Tissue Browning and Muscle Wasting in Infantile Nephropathic Cystinosis-Associated Cachexia"** published July 2021 in *Cells Journal*.

Since 2010, CRF has funded nearly $2 million to Dr. Mak for this important research.

Read full articles on our website:

WWW.CYSTINOSISRESEARCH.ORG/PUBLISHED-STUDIES

CRF RESEARCH

GRANT AWARDS

SPRING 2021 $612,243

RESEARCH GRANTS

Francesco Bellomo, PhD, *Principal Investigator*
Francesco Emma, MD, *Co-Principal Investigator*
Bambino Gesù Children's Hospital, Rome, Italy

"Vanin-1 as therapeutic target in nephropathic cystinosis"

$181,874 TWO-YEAR STUDY

Bruno Vogt, MD, *Principal Investigator*
Daniel Pouly, PhD, *Co-Principal Investigator*
University Hospital of Bern, Bern, Switzerland

"Early events of cystinosis pathogenesis: pro-apoptotic signals and mRNA translation"

$196,090 ONE-YEAR STUDY

EQUIPMENT GRANT

Sergio Catz, PhD
The Scripps Research Institute, La Jolla, California

Celldiscoverer CD7 Zeiss Microscope

$234,279 PURCHASE TOTAL

SEE SPRING 2021
LAY ABSTRACTS
STARTING ON NEXT PAGE

Vanin-1 as therapeutic target in nephropathic cystinosis

Francesco Bellomo, PhD, *Principal Investigator*
Francesco Emma, MD, *Co-Principal Investigator*
BAMBINO GESÙ CHILDREN'S HOSPITAL, ROME, ITALY

OBJECTIVE/RATIONALE:

The metabolism of coenzyme A is essential in the synthesis and oxidation of fatty acids. A key enzyme of this metabolic pathway is represented by vanin-1, which catalyze a reaction that produces endogenous cysteamine. We assume that positive modulation of vanin-1 by agonist drugs or by ketogenic diet could have therapeutic effects whereas the creation of a mouse model knockout for *CTNS* and *Vnn1* genes could generate a model of cystinosis with a more severe and/or early phenotype.

PROJECT DESCRIPTION:

We will create double knockout mice for the *CTNS* and *Vnn1* genes and verify if the absence of the vanin-1 protein abolishes the effects of ketogenic diet. In parallel, we propose to modulate *VNN1* expression by molecular and pharmacological approaches in cystinotic human proximal tubular cells and verify its effects on biological processes that are altered in these cells. Finally, should we identify one and possibly two molecules that are effective *in vitro* we will perform a small pilot study in *CTNS-/-* and double knockout mice.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

We speculated that increased synthesis of endogenous cysteamine is in part responsible for the improvement of the renal phenotype observed in cystinotic mice treated with ketogenic diet. Confirmation of this hypothesis, supported by several experimental evidences, would allow to consider new therapeutic approaches that may be better tolerated by patients.

ANTICIPATED OUTCOME:

The therapeutic power of diet is often underestimated because biochemical processes and metabolic impact are not always predictable. However, our recent studies have shown that ketogenic diet significantly improves the renal phenotype of mouse and rat models of cystinosis, and albeit numerous metabolic pathways are modulated by this diet, the specific reaction catalyzed by vanin-1 has likely a crucial role in mitigating the evolution of kidney disease.

Early events of cystinosis pathogenesis: pro-apoptotic signals and mRNA translation

Bruno Vogt, MD, *Principal Investigator*
Daniel Pouly, PhD, *Co-Principal Investigator*
UNIVERSITY HOSPITAL OF BERN, BERN, SWITZERLAND

OBJECTIVE/RATIONALE:

This project is the continuation of work done in Corinne Antignac's laboratory to understand the role of cystinosin in mTORC1 signaling and mRNA translation. In mouse renal proximal tubular cells (mRPTEC) showing cystine accumulation but no other pathophysiological features, thus taken as an early disease model, we found a transcriptionally up-regulated pro-apoptotic gene similarly to our previous unpublished RNA-seq screenings in mouse kidneys. Second, a translatome screening identified mRNAs related to mitochondria and apoptosis whose translation is increased or decreased after amino-acid withdrawal and reintroduction in cystinotic cells.

PROJECT DESCRIPTION:

The first aim is to study the pro-apoptotic up-regulated transcript that has not previously been reported in cystinosis. We will first determine whether this pro-apoptotic signal can be reduced by cysteamine treatment and study its known molecular cascade partners in our cells. We will also take advantage of knock-in cell lines with nephropathic and non-nephropathic mutations to study the link between this up-regulated transcript and disease severity. The second aim is to study the identified nuclear genome-encoded mitochondrial component whose translation is reduced in cystinotic cells after amino acids withdrawal and reintroduction. We will challenge the cells with repeated cycles of amino acids deprivation and refeeding with all /all but cysteine/ cysteine supplement/ with and without cysteamine to determine whether the observed mRNA translation defect is further increased or corrected, respectively.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

The vision of the lysosome has recently evolved from a passive degradation endpoint to an active player in nutrient availability signaling. We propose the idea that the adaptation to alternation between high and low nutrient availability phases, especially amino acids, may be a metabolic challenge for patient's proximal tubule cells. If the identified targets of both specific aims turn out to have a link with this process, they could be a first step in the elaboration of new therapeutic strategies.

ANTICIPATED OUTCOME:

With this work, we expect to gain insight in the early aspects of cystinosis pathophysiology. The pro-apoptotic transcript may reveal interesting events related to apoptosis already taking place in cells that do not appear morphologically affected by the absence of cystinosin. In the second aim we expect to find out whether cystine accumulation and/or impaired participation of cystinosin in mTORC1 signaling can impact mitochondrial health and apoptosis with altered translation of the identified mRNAs targets upon nutrient availability changes.

CALL FOR FALL 2021 GRANT APPLICATIONS

When Nancy and Jeff Stack established the Cystinosis Research Foundation in 2003, they were committed to aggressively funding cystinosis research to ensure the development of new and improved therapies and a cure for cystinosis. But never in their wildest dreams could they have imagined what has been accomplished in 18 short years. Since its inception, CRF has funded 207 multi-year research studies in 12 countries. Our researchers have published 92 articles in prestigious journals as a result of CRF funding. Every dollar donated goes directly to support cystinosis research.

The goal of CRF is to accelerate promising cystinosis research toward clinical trials. To that end, CRF prioritizes research that will lead to better treatments and a cure for cystinosis. CRF issues grants for bench, clinical and translational research, with a strong emphasis on translational and clinical research. CRF is interested in supporting new investigators and encourages them to apply either as research fellows or investigators.

In September, CRF announced $2.5 million was available for the fall 2021 call for research and fellowship applications. The grant awards will be announced at the end of December 2021.

In spring 2021, CRF issued three grants; two new research grants and one equipment grant which total $612,243 that will bring us closer to better treatments and a cure. All research applications received by CRF are evaluated by CRF's Scientific Review Board (SRB), composed of the leading international experts in the field of cystinosis.

The SRB provides independent, objective reviews and recommendations for each research proposal submitted based on the NIH scale of standards. Additionally, the SRB follows grant review guidelines established by CRF and advises the foundation on the scientific merits of each proposal.

In 2021, CRF updated the registry questionnaire to include questions that are relevant to recent scientific advancements, new medications and patient care. CRF partnered with CoRDS *(Coordination of Rare Diseases at Sanford)* creating a new Cure Cystinosis International Registry, the only international cystinosis patient registry in the world. The site includes a professional Research Portal so that researchers and scientists who register can access and view de-identified, aggregate cystinosis patient information. The registry will connect all of the stakeholders in the cystinosis community – the scientists, researchers, clinicians, pharmaceutical companies, patients, and families – and provide them with resources that have never been available in one place before, all to accelerate better patient care.

Cure Cystinosis
International Registry

We encourage every family and patient to enroll today. Your information is essential to advancing cystinosis research. If you have already enrolled, thank you!

Visit the CRF website to learn more about CCIR and enroll.

WWW.CYSTINOSISRESEARCH.ORG/CURE-CYSTINOSIS-INTERNATIONAL-REGISTRY

WWW.CYSTINOSISRESEARCH.ORG/APPLY-FOR-RESEARCH-GRANT

CRF is excited about the future of cystinosis research and is grateful to researchers for their interest in the cystinosis community. We look forward to working together to find better treatments and a cure for cystinosis.

CYSTINOSIS RESEARCH FOUNDATION

SCIENTIFIC REVIEW BOARD

The CRF Scientific Review Board (SRB) is composed of leading cystinosis scientists, researchers and clinicians from around the world. We are indebted to our Scientific Review Board members for their leadership, guidance and commitment to improving the lives of our adults and children with cystinosis. Thank you!

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THANK YOU FOR YOUR DEDICATION TO THE GLOBAL CYSTINOSIS COMMUNITY

WELCOME

New Board Member

Angela Kirchhof

DENVER, COLORADO

My four-year-old daughter, Hayden, was diagnosed with cystinosis at 14 months old. Back when we first Googled cystinosis, I never could have imagined that our life would feel so normal and routine a short three years later. While managing cystinosis is a job that everyone in our family shares – even Hayden's two-year-old sister knows when to grab a bowl because Hayden is going to get sick – it's one that operates in the background most days. My husband, Nick, and I didn't expect to develop the medical vocabulary that we have or take such a keen interest in the advances in medical technology. But today, we're always sharing with each other news stories that we find about other families living through similar challenges. Nick plays an active role as the Colorado ambassador for NORD – often reaching out to newly diagnosed families outside of the cystinosis community to make a connection. We've seen firsthand how much of a critical difference it makes when a family feels supported by a community that can empathize with their experiences, and we look forward to paying that forward.

I have more than six years of experience in roles where I've built up new teams and transformed them to highly functional ones. These are all skills that will be valuable to draw upon in my position as a board member. I'm able to take input from many different sources and turn it into a vision and a rough roadmap for how we can achieve that vision. I love building new relationships and am excited by the opportunity to build new ones with families and researchers dedicated to improving the lives of those with cystinosis.

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See page 67 for Fundraising Events in Honor of Hadley.

By Marcu Alexander, Stella and Hadley's mom
BOISE, IDAHO

It's hard to believe that 2021 is nearing an end and soon we'll be ushering in a new year. I'm hopeful 2022 will be easier than the past two years we've experienced due to the pandemic. I'm ready for life to safely resume and for my kids to enjoy their childhoods without living in fear of a loved one or themselves getting sick. It's hard enough to balance life when you have a child with cystinosis but adding a global pandemic on top of that is a whole other story. It's certainly taken its toll on my mental and emotional state, but I choose to power on and make the most out of a difficult situation. One of the hardest things my family endured was the physical distance from friends and family. Remote school and work along with the cancellation of vacations, events, and virtually everything else we had planned was disappointing, but nothing hurt as badly as being apart from loved ones.

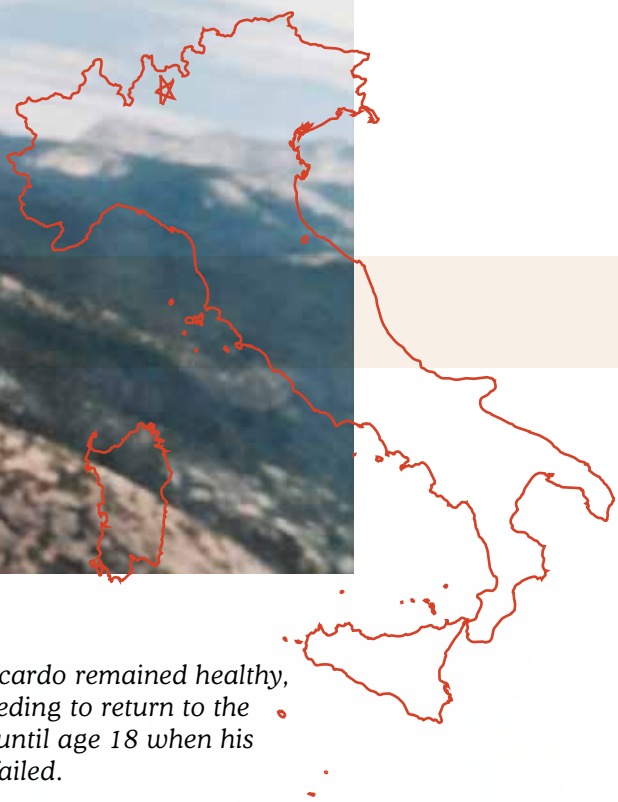
When the world briefly opened this spring and summer, we took full advantage by re-connecting with friends and family we haven't been able to spend much time with during the past year. Ben, Stella, Hadley, and I never realized how much we took spending time with others for granted. We packed in a lot of adventures during the girls' summer break. We started off by celebrating the end of school with a camping trip to Lowman, Idaho with some friends. The weather was perfect, and it felt nice to be out in the wilderness without electricity or access to devices. We enjoyed going on nature walks and chatting by the campfire with coffee in the morning and a cocktail at night. The kids had fun playing games, getting into water fights, and making cookies on a solar stove.

In July we spent only one weekend at home and the rest of the time we were off having fun. We enjoyed the 4th of July in one of our favorite places to visit, McCall, Idaho. McCall is situated on Payette Lake where days are spent relaxing and swimming in the lake. Our friends have a cabin on the lake and hosted an unforgettable 4th of July celebration which included an epic water parade on floaties, face painting, games, and fireworks!

We had the pleasure of returning to McCall toward the end of July which allowed us a chance to spend time with the Partington family. We've made it an annual summer tradition to visit them in McCall since we moved to Idaho eight years ago. We first met the Partington family when Hadley was diagnosed with cystinosis at 18 months. Teresa and Kevin provided much needed emotional support as well as tips and tricks that made caring for a newly diagnosed child with cystinosis easier. Jenna and Patrick have always been like a big brother and sister to Stella and Hadley, and my girls adore them. Cystinosis may have been what brought us together, but a true friendship was formed. It doesn't hurt that Teresa is originally from Idaho and we share many common interests! We hadn't seen them since the 2019 Day of Hope family conference, so the reunion was incredibly fulfilling.

Now that the girls are back to school and the pandemic has reared its ugly head again, I've had the chance to reflect on the wonderful reprieve we had over the summer. I'm grateful for all the fun we had and the new memories we made. There is no greater gift in life than the relationships we cultivate with others. It reminds me of a quote by one of my heroes, Brené Brown, *"Connection is why we're here; it is what gives purpose and meaning to our lives."*

RICCARDO FOGLIADA



associazione cistinosi

Italian Association for Cystinosis

This is the story of Riccardo, a 27-year-old Italian man with cystinosis. The diagnosis was made when he was 18 months old because, at that time here in Italy, the illness was little known.

Thanks to our relatives in California, we learned about Dr. Jerry Schneider and his many patients. We went to San Diego in 1996 when Riccardo was two-and-a-half years old. There, we were guests and living examples at a lecture on cystinosis given by Dr. Schneider at the University of California San Diego. With us was a family from Orange County with their beautiful little blonde daughter, Natalie.

After having met with Dr. Schneider in San Diego, we learned to optimize Riccardo's care. Riccardo remained healthy, never needing to return to the hospital until age 18 when his kidneys failed.

We decided that I would donate a kidney to him. At age 19, he received a living donor transplant but unfortunately, two days later, due to a blood clot in the renal vein, the kidney had to be removed and he had to return to dialysis. At the time, he was a student at the University of Milan, and he was on dialysis for four years. We never received any notification about a cadaver transplant, so his father decided to donate one of his kidneys to Riccardo. The second transplant took place in 2017. This time, all went well and to date, Riccardo has been healthy and lives the normal life of a student and a young man of his age, (aside from the numerous medications he must take and the strict schedule he must follow).

During numerous trips to California, we got to know the very dear Mrs. Jean Hotz and her foundation, which I believe

By Mara Mazzina, mother of Riccardo Fogliada

GORDONA, ITALY

was the first founded in the United States. We attended and participated in numerous conventions and met and got to know many families. Even though the first convention we attended was a shock to us, over time we learned much from both doctors and the other participating families, and we always received help and support.

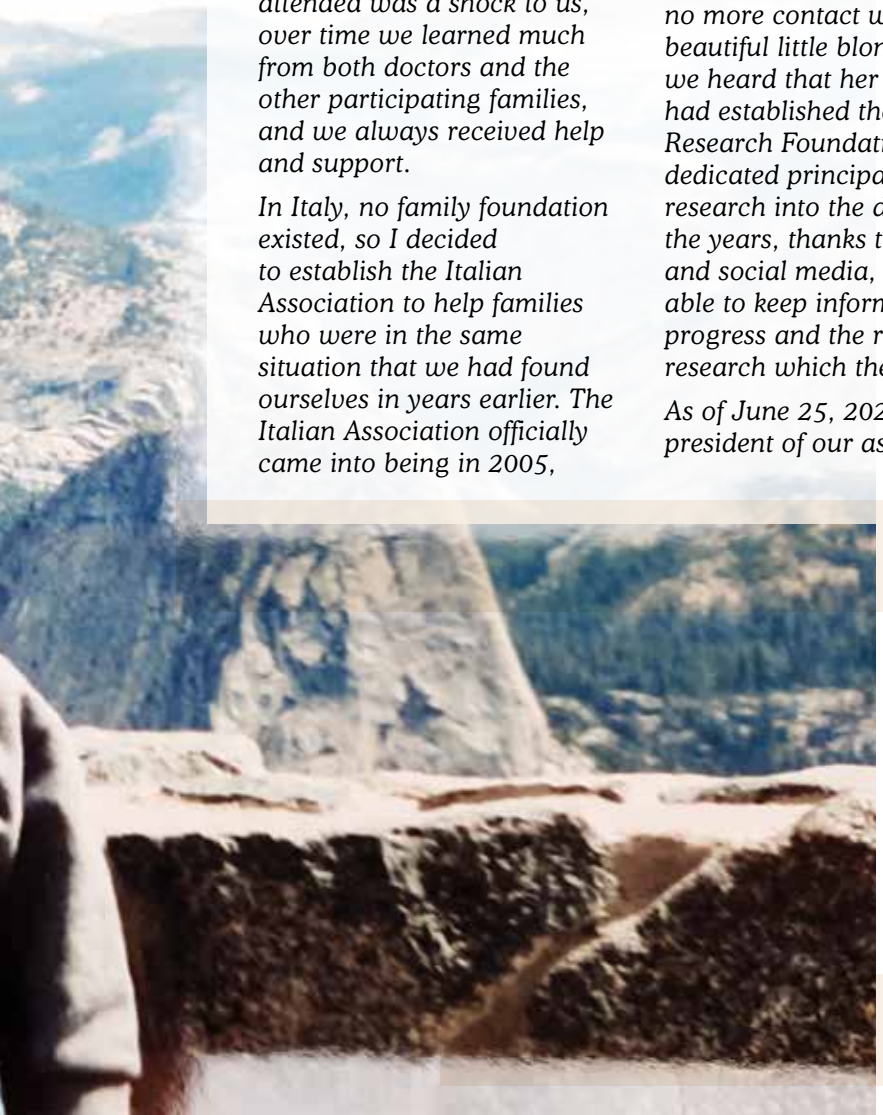
In Italy, no family foundation existed, so I decided to establish the Italian Association to help families who were in the same situation that we had found ourselves in years earlier. The Italian Association officially came into being in 2005, but had been active since 2000. We organized two international conventions in Italy in 2000 and 2010.

Unfortunately, after that first visit to San Diego, we had no more contact with the beautiful little blonde girl until we heard that her parents had established the Cystinosis Research Foundation dedicated principally to research into the disease. Over the years, thanks to technology and social media, we were able to keep informed of their progress and the results of the research which they financed.

As of June 25, 2021, the new president of our association is Mrs. Laura Morocutti, mother of a 16-year-old girl with cystinosis. We wish every success and thank her for her involvement.

And to the Stack family and their foundation, to whom the entire cystinosis community must say a huge "Thank You" for their involvement and dedication to finding a cure and to fulfilling the wish

"to have my disease go away forever."



TRUE FRIENDSHIP IS RARE

By Jill Emerson, Brooke's mom
HAMMONTON, NEW JERSEY

"Friendship... is born at the moment when one man says to another, 'What! You too? I thought that no one but myself.'"

C. S. LEWIS

This quote perfectly sums up how it feels when, as a parent of a child with a rare disease, you find a true friend who is also a parent to a child with that same disease. And what is even more rare than that, is when that friend's child is close in age to yours, and you live in close proximity to each other!

That is how I feel about my friendship with Leanne Pollock, whom I met at a cystinosis conference years ago. Leanne's daughter, Amelia, and my daughter, Brooke, are close in age, which has certainly made our bond stronger. And the fact that the Pollocks live relatively close to us makes it all the more special!

In the winter of 2020, Clay (my husband), Brooke and I were able to plan a long weekend getaway with the Pollock family – James, Leanne, Amelia, and Ellie. Having found a beautiful home to rent for the weekend, we were excited to get Amelia and Brooke together (and Ellie, Amelia's baby sister, too!). The girls were able to play in the big backyard, and we were greeted with a snowstorm right before arrival, which was perfect for the girls' snow angels.

Clay and I feel lucky to have been able to spend that time with the Pollocks, especially since only two months later, COVID hit. Since then, it's been difficult to plan any outings given the virus; but it hasn't been for lack of trying! So, we were thrilled when we were able to get together for a beach day this August before the girls went back to school. Leanne, Amelia and Ellie joined us for a day filled with sand, sun, laughter and quesadillas.

The girls had so much fun playing in the sand, putting their feet in the ocean, and reacquainting themselves with each other. Nineteen months is a long time when you're a kid, and the girls have grown and changed so much in that time. Amelia and Brooke couldn't be more different in personality; Amelia is outgoing, Brooke reserved. But they complemented each other so well, and really hit it off. After

REACHING MILESTONES TOGETHER

the day at the beach, we headed back to the house and the girls played nicely with each other on their own, giving Leanne and I some moments together. A day trip definitely wasn't long enough though, and I know Brooke is looking forward to seeing her friend Amelia again as soon as possible. Immediately upon the Pollock's departure that day, Brooke was inquiring when the girls' next playdate could be!

Just like we adults benefit from and need friends who understand what life as a cystinosis parent is like, I believe our children can benefit from friends within the community as well. That is why attending the Day of Hope Conference has been so vital and special for Clay, Brooke and me. And it becomes more evident as Brooke gets older that she will benefit from having friends within the community who understand her.

While we don't live around the corner from each other, we do live close enough for day trips, and as the girls get older, I plan on taking advantage of this proximity to continue nurturing their friendship. I know both Leanne and I are thrilled that Amelia and Brooke have each other to rely on as they get older, to spend time with when things get tough, and to celebrate each other as they reach amazing milestones that both these girls are destined for.

SEE THE MARCH MADNESS CHARITY POOL IN HONOR OF BROOKE ON PAGE 66

living life

By Mark Flerchinger, Tina's dad
CLARKSTON, WASHINGTON

When you meet Tina, you will immediately notice her big beautiful smile. She is very approachable and has a genuine charm that attracts people to her. She really is fun to be around, always has something funny to say, and is quite the conversationalist.

People will often say her mother and I have taught her well. I tell them that I have learned more from her than I will ever be able to teach her. I am always awestruck, but not surprised, when people tell me how Tina inspires them. She takes her struggles in stride, always with a smile and a resolve to keep things positive.

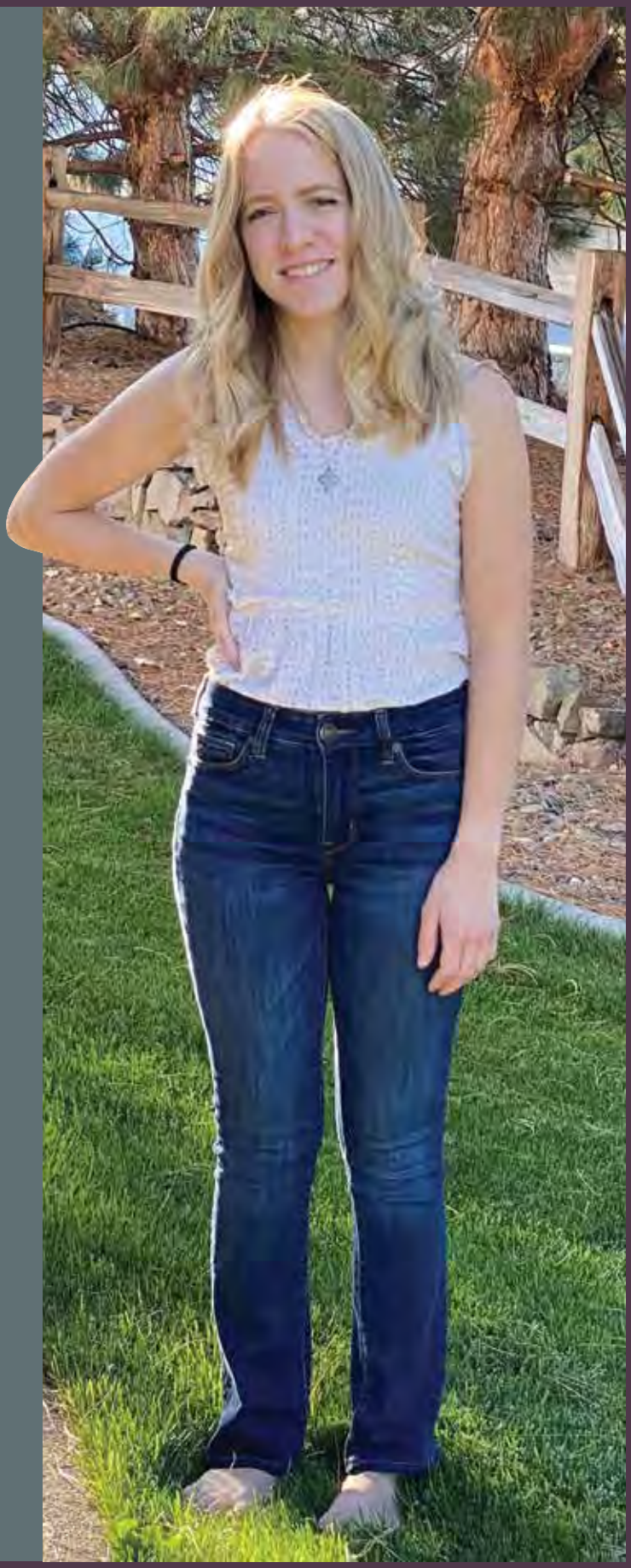
The last two years have been different. During the early days of the pandemic, Tina took up painting with Bob Ross. Her landscape portraits are quite remarkable. We have been the beneficiary of her trying new recipes. She enjoys being in the kitchen – it usually involves her dancing around in her mama's apron listening to 80's music. Tina has the same motto as her Uncle Dick when it comes to food - "Life is too short to eat a bad meal."

"life is too short to eat a bad meal."

Being able to give Tina my kidney was the greatest gift I could have given her. Despite it being an old worn-down kidney, it seems to be doing well in its new home. It is awesome to see Tina thriving. After the kidney transplant two years ago, she made a pact that she was going to "start living life" and she has done just that! She now has her driver's license, works a job at TJ Maxx, and enjoys country swing dancing on the weekends (she got the moves from her mother).

Tina still has days where she is not feeling well. She gets frustrated with her medications, understandably due to the quantity and negative side effects. On these days you will find her napping with her dog, Sam. Since Tina was a little girl Sam has been her protector. It's as though he can feel when she is not well and she finds great comfort with him.

I admire the courage my daughter has shown through her short life. She inspires me every day to be a better version of myself. Don't get me wrong, on the days where she is greatly challenged, she may lose her temper, but I think it's cute. Her older sisters will say she gets away with murder and that I can never say "no" to anything she asks for. This is true, but I am working on it! Tina has so much empathy towards others who suffer from illness. These are the same traits her mother holds. She often worries about her cystinosis friends and prays daily for them. But in the midst of all of this she takes on each and every moment with enthusiasm and joy while holding onto the hope of a brighter future for her cystinosis friends – she is so happy that a cure is on the horizon! I am proud of the young lady that Tina has blossomed into. She is smart and witty, a senior in high school and counting down until she will attend college in Fall 2022. I can't wait to see how her life unfolds and what the future holds for her.



DRIVING HOPE

By Mack Maxwell

BURLESON, TEXAS

> *"I'VE HAD MY TRANSPLANTED KIDNEY FOR OVER 46 YEARS.*

My journey to retirement is almost complete. In August 2021, I told my managers at Tarrant County College (TCC) that I would be retiring on December 31, 2021, at age 57. My career in Information Technology includes 34 cumulative years at TCC and 1.5 years at Sabre Corporation. My retirement package consists of an excellent pension, free health insurance for life, and solid investments. My retirement plans include a lot of travel, playing poker, and being an active member of the Cowtown Corvette Club.

CYSTINOSIS EARLY YEARS

I was born on April 9, 1964. My infant years were normal, except for photophobia and slow growth. It took a long time to determine a correct diagnosis in the early 70s. At age eight, local doctors suspected that I might have cystinosis and this was confirmed by Dr. Harold Harrison in Baltimore, MD.

KIDNEY TRANSPLANT

My biological mother, Lynne Blissit was contacted in 1975 and offered to give me one of her kidneys to save my life. Lynne's kidneys were a perfect match. Fortunately, I only had to go through three weeks of dialysis before my transplant. The kidney transplant was performed on June 10, 1975, by Dr. Paul Peters in Dallas, TX at Parkland Hospital. As of August 2021, I've had my transplanted kidney for over 46 years.

TREATMENT AT THE NATIONAL INSTITUTES OF HEALTH (NIH)

After my transplant, I felt great except for my eyes. For years, I had severe photophobia and eye pain that got worse with age. In June 1999, I discovered there was a cystinosis treatment available for my eyes at the NIH. My first visit to the NIH was in September 1999 at age 35. This was my first time meeting with Dr. Gahl and Dr. Kaiser. Dr. Kaiser gave me the NIH cysteamine eyedrops which instantly eliminated my eye pain and changed my life. One year later, my eyes were completely crystal-free.

During this visit to the NIH, Dr. Gahl put me on cystagon. Taking cystagon was a huge challenge because of the severe side effects. Procysbi® became FDA-approved in 2013. Procysbi® has been much easier for me to tolerate.

A LIFETIME DEDICATED TO PHYSICAL FITNESS IS ONE OF THE REASONS THAT I'M STILL IN GOOD SHAPE WITH CYSTINOSIS AT AGE 57.

CYSTINOSIS CONFERENCES

In August 1999, I went to my first cystinosis conference in San Diego, CA. This was the first time that I had ever met another person with cystinosis. This was the first of many conferences that I would attend in both the United States and Europe. The European conferences have given me the opportunity to visit Amsterdam, Barcelona, Paris, Dublin, Prague, Venice, Manchester, and Scotland. I've had the opportunity to develop a global network of friends because of cystinosis.

Being one of the oldest people with cystinosis, I try to offer some advice. The most important advice that I can give is the importance of exercise. A lifetime dedicated to physical fitness is one of the reasons that I'm still in good shape with cystinosis at age 57. I've had the opportunity to speak about this at conferences several times throughout the years.

LIFE LESSONS FROM PARENTS

I thank my mother for teaching me to have a positive attitude and teaching me to believe that I could accomplish anything I set my mind to. That mindset was ingrained in my brain before I started first grade.

I thank my father for my hard work ethic. I helped him with many projects throughout the years. He grew up in the depression era and knew the value of hard work. He led by example and was one of the hardest-working men that I've ever known.

AFTER DECADES IN INFORMATION TECHNOLOGY, MACK IS SHIFTING INTO RETIREMENT...

A SMOOTH SEA NEVER MADE A SKILLFUL SAILOR

By Carli and Kevin Simpson, Charlie's parents
NEW ORLEANS, LOUISIANA

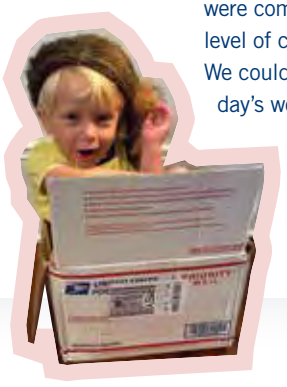
////////////// **CHILDREN'S HOSPITAL NEW ORLEANS** EARNED THEIR STRIPES THIS YEAR //////////////

Almost ten years ago, as newlyweds, we were given the opportunity to move to New Orleans, a unique American city where we could do a "stint" for a few years. We're now approaching the end of a decade in this treasured place, where we bore two children and have banked so many memories.

But it's not the culture or cuisine that have given us our most meaningful "Big Easy" experiences. Children's Hospital New Orleans was where our little boy's life was likely saved when a mysterious illness had him (and us) down for the count. Unable to keep anything down and so dehydrated that sticking a vein was almost impossible, our infant son might not have survived had the staff not been as mindful, well resourced, and present as they were.

It's never as seamless as it looks. A hospital staff has issues in common with any other workforce; it's comprised of human beings who are subject to the same laws of nature, the same accidents, inconveniences, and conflicting work-life priorities we all face. In this particular place, the occasional hurricane can impact all those variables; the most dedicated nurse cannot get to work if his car is low on fuel and the gas stations have no power; the best phlebotomist is challenged to focus on finding a vein if her child's bedroom has a tree through the window; and the most seasoned physician cannot magically appear at the hospital if the streets are impassable.

Hurricane Ida hit New Orleans on August 29, 2021, so it's fresh in our minds as we write this article. She was relentless -- the first storm forcing us to evacuate since moving here. We'd been resistant to leave home because Charlie's routine is so critical to his health, and travel tends to throw that routine off, sometimes badly. It's also tricky to transport his many medicines and supplies and administer them on the road, especially when leaving for an undetermined amount of time. But evacuating was unavoidable. Ida's powerful winds took out electricity to the entire metro area indefinitely. Fuel shortages threatened essential needs, and even cell phone and 911 services were down for days.



Yet Children's Hospital stayed open. And Charlie's devoted nephrologist – who always makes herself available to keep Charlie stable and thriving, all while raising two young children of her own - actually stayed in town to care for patients.

This made us contemplate how much more grueling the discovery of early cystinosis in our child might have been had it happened at a hospital seriously impacted by the COVID surge and in the aftermath of a major hurricane. The experience was chaotic and terrifying in relative peacetime. Yet in this "Ida" moment, Children's Hospital overcame those hazards. Its leaders made hard decisions and likely called in every favor to get critical resources and staff on-site. At a time when the world seems like it's spinning out of control, imagine if your job was to emerge from that madness and come to the rescue of distressed children, and their distraught parents, multiple times every single day.

It's unlikely that a cystinosis case was present at the hospital during Ida but it's certain that equally severe cases were coming in the door – and were handled with the same level of competence and compassion that we experienced. We could ask them what it's like, but we guess it's all in a day's work. Even so, we feel immense gratitude for those lifesaving professionals who, without reliable access to even basic societal needs and under pandemic conditions, somehow navigate the devastation to accomplish their work.


HURRICANE IDA WAS THE FIRST STORM FORCING US TO EVACUATE SINCE MOVING HERE.

We share the same pain — we share the same laugh.

By Asim Bukhari, Rayan's father
DHAHRAN, SAUDI ARABIA

My wife (Sadia) and I have been living in Saudi Arabia for a decade now. After having two beautiful daughters (Faria & Zaria) in our family, life was just about perfect when we had another kid. My son, Rayan was born in November 2015. It was all good until at age eight-months he was dehydrated and lost his charm within a few days. It was an early sign of cystinosis. To be honest, it might be difficult for most pediatric specialists or nephrologists to identify the disease at first glance and with few symptoms. Rayan was finally diagnosed with cystinosis early this year (January 2021) after having a genetic test.

Unawareness is a great blessing, but in some cases knowing is an even greater blessing, so this is where our journey begins.

It's an honor to speak to the world through this magazine, but when I was first asked to write something for the CRF Cystinosis Magazine, I really struggled to figure out what I wanted to say or how I felt. I had to represent all other families around the world who haven't had a chance to write their story.

Like all other families, at diagnosis, we had no information about the disease and when we googled it, we were heartbroken. We cried a lot, thinking that this was the end of the world – will our son never live a normal life?

Yes, we were very depressed but never hopeless. We always had a belief that this is something we never wished for, but Almighty Allah will guide us to the right path. Eventually, we received an instant reply from Dr. Stéphanie Cherqui from my email which opened the door of guidance for us. She introduced us to her team and then lead the way to Mrs. Nancy Stack and CRF, doctors, specialists and the cystinosis community. Since then, the way Mrs. Stack took care of us during our anxieties - we have no words to describe. I would also like to especially thank Jordan Janz and his mom, Barb, for helping us recover from the initial trauma. It may sound unreal, but indeed all of these people are always with us whenever we need them. They are special to us.

How has our life changed after the diagnosis...? (That is another story...). Life is precious and living life with any disease brings value to it. Rayan is taking multiple medicines for his thyroid, kidney, and other complications so that his body can function properly. He is cooperative so far in terms of taking medicines on time which makes things easy for us.

Rayan has big dreams; he wants to be a pilot. I don't know if the obstacles he has now will ever let him live a healthy childhood (the kind we had) but I am sure he will grow up to become one; "God willing."

We have also learned that difficulties in life don't mean the end of the world, but rather the difficulties make us stronger than before. Now, we enjoy all the joys and mischiefs of our son - we are prepared to challenge the disease.

As said by one physician, there is so much that you know about this disease but there is still enough that you don't know. I will close my words by narrating a verse from the Holy Quran which has been mentioned in connection with the right to life. Almighty Allah has said: **"And whoever saves a life it is as though he had saved the lives of all mankind"** – and CRF is doing exactly that, irrespective of nationality, race or color. They are doing excellent work by bringing the world together. We share the same pain; we share the same laughter. Thank you.

12 is a perfect number

By Jackson Blum-Lang and
Jean Blum, Jackson's mom
SCOTTSDALE, ARIZONA

Hi, I'm Jackson, and today is my birthday!!! I am officially 12 years old. Did you know that 12 is a perfect number? Don't worry, I didn't know that either. My Mom told me.

I wonder what it means to be the perfect age. It must mean that good things are going to happen, right? But what good things? Can I decide? I say yes!

THESE ARE THINGS THAT I WOULD LIKE TO HAPPEN TO ME WHILE I'M THE PERFECT AGE:

- I grow a little bit in all 12 months of being 12
- I go 12 straight days without a tummy ache from my meds
- My breath smells like nothing instead of my medicine, every waking hour (about 12 hours a day)
- A bunch more grownups get cured of cystinosis so the minimum age for the treatment goes down to 12
- I get an iPhone 12
- I take medicine every 12 hours instead of every 2-4
- I need eye drops every 12 days instead of every day
- I find 12,000 Robux in my account
- My Mom gets to sit and breathe for 12 straight minutes a day instead of constantly doing med prep, running me around to appointments and activities, and working

Sorry, I walked away from my computer for a minute, and I found Mom typing that last bullet point when I came back. I'll allow it to stay.

I guess these aren't the typical 12-year-old wishes, are they? Don't feel bad for me, though. I find time to do lots of fun stuff. I take a trampoline class twice a week. I code video games. I play video games and I watch YouTube videos of everything you can think of. I'm great at anything science-related. I make YouTube videos (channel: DoubleJdude). I hang out with my friends, playing video games, swimming, biking or do whatever else we can think of. I'm really good at go-kart racing and I'm a pro at laser tag. I also love playing with my crazy French Bulldog, Henry.

Thanks for listening. Mom will take over from here.

Hi! Jean here.

This feels a bit like "…and now a word from our sponsors," where everyone hits mute or turns the channel.

I do want to say a few words, however, beyond asking for 12 minutes to breathe every day.

Jackson's journey has been very similar to others traveling down the cystinosis road, so I won't talk about the early days of diagnosis, vomiting, not eating, vomiting, medications, vomiting, no sleep, vomiting, and so on. I do, however, want to talk about gratitude, because I have a lot of it.

I am extremely grateful for the Cystinosis Research Foundation, and its laser focus on improved treatments, and ultimately the cure. I have never-ending gratitude for everyone who does fundraising events, and/or digs deep into their pockets to support the CRF's focus. I have so much admiration for all the CRF researchers and doctors who are dedicated to our small community. It is because of everyone I just mentioned that Jackson and I can live without fear.

When Jackson tells me that he is worried about his future health, I can confidently tell him that he is going to be just fine. When I read about all the exciting advancements that are happening, I must hold the tears back *(sometimes unsuccessfully).*

The future is bright indeed!

Moa

By Pontus and Frida Brolund, Moa's parents

KARLSTAD, SWEDEN

Our journey started some 28 years ago when we met each other. We are Moa's parents, Frida and Pontus. We met in a basketball gym in a small town named Karlstad in Sweden. We became parents early, when we were 24 years old. We had a beautiful girl named Hugo or I mean, Moa!

Everything was great until Moa started eating baby food at eight months old. She would not eat anything, and could go several days eating only half of a meatball and a slice of cucumber. We were thrilled when one day she would eat half of a banana. Then, she would be back to eating almost nothing for a few days. At 14 months, she received the diagnosis of cystinosis. When we look back, we are happy that we were so young because, as young parents, we were somewhat naïve; we didn't see all the problems that were ahead of us.

There are several families in Sweden with cystinosis. We attended a meeting in Sweden with other families and we heard a rumor about the Cystinosis Research Foundation and their annual conference but we got spooked thinking about all of the sick kids and meeting with their parents. We were afraid to research more about CRF because of our earlier experiences. We found comfort in the Kühnel family who live in Sweden and who have a child one year older than ours. We traveled many times abroad in Europe, worked full-time and lived a normal life.

Moa is a strong and clever girl and she is doing great.

Then one day when we were in Gothenburg for our yearly check-up, we met up with the Kühnel family. They had just arrived home from Los Angeles from the CRF Day of Hope family conference and Natalie's Wish event. They were so thrilled and happy and told us "You must go there!" We were still negative about the idea; we said it can't be that awesome, people can't be that nice and helpful and no, the cure is not that close.

But then, we made our first trip to the U.S.A. in 2016. It was so perfect and we met so many nice and beautiful people. We learned so much about everything and Stéphanie Cherqui talked to us about her work and about a cure. We couldn't believe the research progress but now, three people have been transplanted and are cured. It was the best trip ever and when we got home, we were positive about Moa's future and cystinosis. The other big change was Moa started Procysbi® and that changed our life for good - as a family we finally got to sleep.

The one thing we were worried about was when she turned 18 years old because we had to leave the pediatric care and find doctors who treated adults with cystinosis. We were worried that as her parents, we would not be able to be with her at her doctor appointments. But Moa is a strong and clever girl and she is doing everything by herself and she is doing great.

Now Moa is 22 years old and living in her own apartment with her dog, Bamse. Last year her kidney function started to fail and she is waiting for a transplant operation in October. Pontus, her father, will be donating one of his kidneys to her. Now we just have to be patient and wait for the surgery and hope that everything will go well. Then Moa wants to have the stem cell transplant (the cure) and be healthy and live a long life. Moa is now studying to become a nurse and she will be the best nurse ever.

It was the best trip ever and when we got home, we were positive about Moa's future and cystinosis.

One on One

with THE FENN FAMILY

By Chris Fenn, Ethan's dad

RUTHERFORD, NEW SOUTH WALES, AUSTRALIA

My name is Chris Fenn, I am the father of an amazing little boy with a terrible genetic condition called cystinosis.

I decided to write a small update on Ethan and his endeavour to cope with his condition. After considering the best way to approach this, I figured I'd get some thoughts from his family. One-by-one, I spoke to each family member. I explained I would ask them something and record their answer. That there were no right or wrong ways to answer. To just tell me some words that come to mind, speak out their feelings, anything they wanted. I asked each person, *"What are your thoughts about Ethan and cystinosis, what do you think about the future, the research, and the hope that you have?"*

HERE ARE THEIR ANSWERS:

ALIZABETH *(Big sister), 12 years old:* "I have a lot of hope for the cure and that Ethan will get his wish to be normal."

PHOENIX *(Big brother), 11 years old (today):* "I don't really know. Um, I hope that there's a cure soon, maybe in the next few years."

JAXEN *(Big brother), 9 years old:* "Um, –deep thought– I hope he gets a cure –deep thought– I don't know. Happy and sad." {Question from me – Why happy and sad?} "Happy because I am confident there will be a cure. Sad because Ethan shouldn't have to go through that. No one in the world should." {Question from me – Why are you confident about the cure?} "Because they have been working really hard." {Question from me – Who has been working hard and how do you know?} "Americans, like, trying and doing a lot of research and putting a lot of stuff together to find a cure. Lots and lots of money to find a cure for cystinosis. Because I saw them when we went to America to the conference, everyone was working together and giving so much money."

MORGAN *(Little brother), 6 years old:* "Um, for Ethan, and his cystinosis, I feel bad because he has to take medicine every six hours and I don't."

AMANDA *(Mum), 37 years old:* "Resilience. Hopeful that he will be cured. Um, can't wait for the day that he says that he used to have cystinosis. Being able to prove people wrong. {Question from me – What do you mean by "prove people wrong"} "To say that he did it, he conquered, he kicked cystinosis' arse. He survived without a hint of bitterness, with a winning attitude and never once contemplated defeat."

ETHAN *(Has cystinosis), 8 years old:* - long thought – "I don't know. I'm sorry, I don't know." {Question from me – It's okay mate, there is no right or wrong answer, do you think you could tell me why you are finding it hard to answer?} "I just don't like thinking about it and it's hard to say what I feel. I guess, I would want to say thank you to the doctors that are making a cure and to all the people that help me go to America." {Question from me – To the cystinosis conference?}

"Yeah, that's where the cure doctors are."

AS FOR ME, when I think of Ethan and cystinosis, one word comes to mind. Hope.

LAUGHTER *is the* BEST MEDICINE

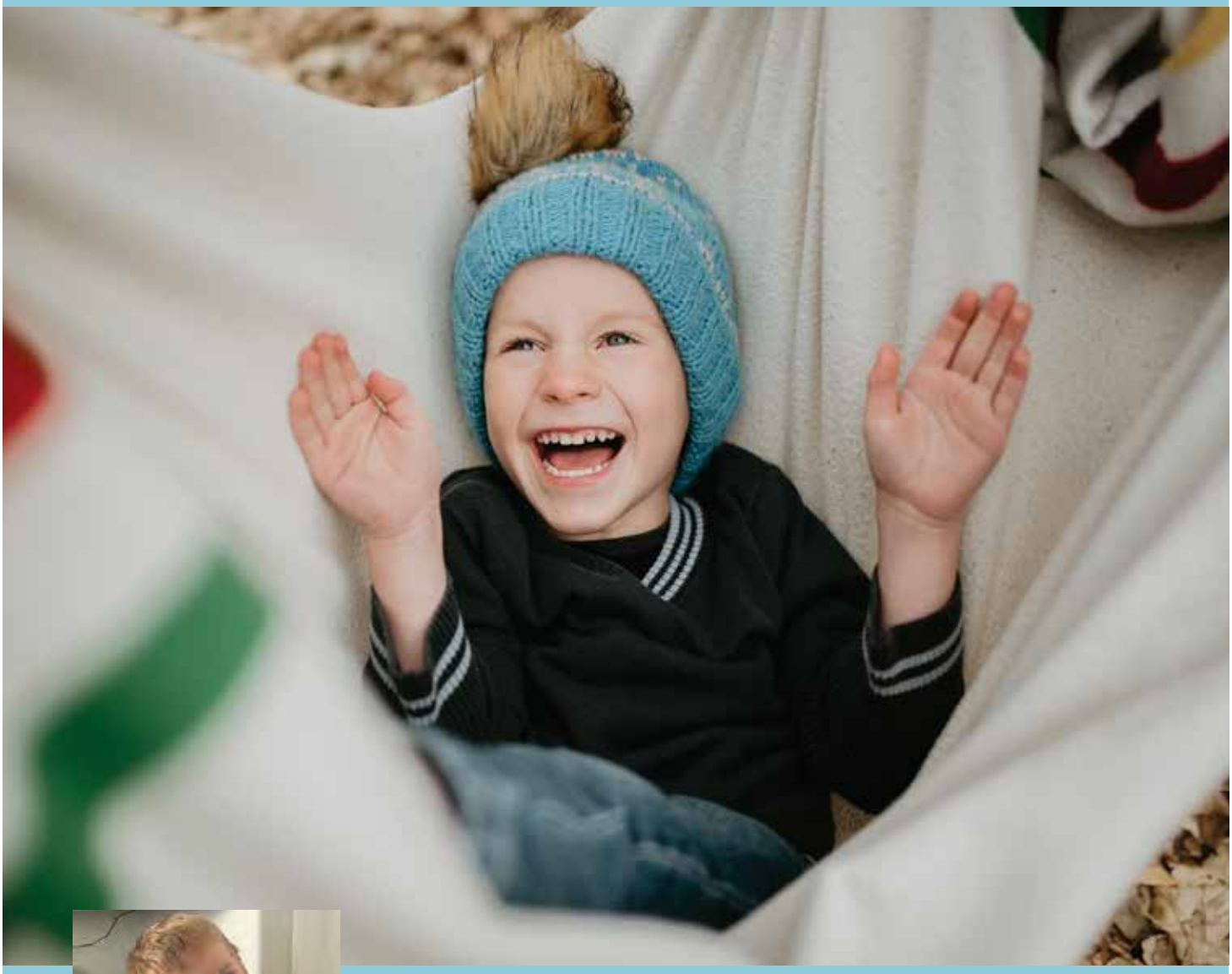
By Leah Klaassen, James' mom
WARMAN, SASKATCHEWAN, CANADA

James was diagnosed with cystinosis four years ago this last summer, and while those memories are fresh in our minds, it also feels like forever ago. We were incredibly lucky in that James was diagnosed very quickly once we went into the hospital the first time. The last four years have been hard, but I am happy to say that there are more ups than downs these days. We've been fortunate that James hardly ever gets sick from his medications, and is as healthy as he can be. He celebrated his fifth birthday in July and started Kindergarten in September. You can usually find him running, dancing, climbing trees or digging up "dinosaur bones" in the backyard. He makes friends fast with his chatty nature and informed us the day before Kindergarten that he planned on being the clown when he arrived. Making people laugh might be his favorite thing.

After James' diagnosis, we found it incredibly difficult to decide whether or not to grow our family. James was an only child, and we didn't know what to do next. We had always planned on more kids, but this seemed to change our plans. We did take our time with that decision and ultimately decided to have another baby. James' little sister, Maya, was born in January 2021, and she has been a perfect addition to the family. James adores her. He loves her tiny toes and making her giggle with his sweet moves and silly faces.

Managing cystinosis is mentally and physically draining. There is always a worry in the back of our minds as to when things will get worse or something new will come up. James is the only one in Saskatchewan with cystinosis, so we are grateful to be a part of a tight-knit community where the support has been incredible. We have learned to lean into that, and it has made us better and stronger. We have been able to host some smaller fundraisers during the pandemic but look forward to doing our annual golf tournament again next year. We also do an annual crokicurl tournament in wintertime (think curling meets crokinole). The first year we did the crokicurl tournament it was -50 degrees Celcius, but that didn't stop us Canadian prairie folks from gathering outside to raise money for a good cause. We have some very enthusiastic supporters!

Throughout this journey, we have learned a lot of things. We've learned that you can go through something so hard, and still be thankful for the path and the journey it's put you on. You can feel pain and grief and joy and thankfulness, simultaneously. You can wish something to go away, or that it wasn't a part of your story, but also recognize that you have learned so much from it and that you don't know if you would be the same person without that hard thing. We've learned that strength is not something you always feel, but that you practice. Some days you feel



James with his little sister Maya.

strong, and some days you fall to the ground, and both are okay. We have found that the more open we are about our struggles, the more support and love we get, and we need that to keep going.

The cystinosis families and CRF have welcomed us since the beginning. It was at our first Day of Hope conference in 2018 where we finally felt hopeful for James' future. We left that weekend feeling lighter and happier. This community of families sharing lived experiences and supporting one another has become an integral part of how we care for James, and ourselves.

The Cystinosis Research Foundation has offered us hope when we couldn't find it on our own. Thank you to all of the donors who continue to support the CRF, and to the researchers who never quit. We are so grateful to all of you.

Smiles of Hope

By Ava Long, Lola's sister

CHASKA, MINNESOTA

When Lola was first hospitalized, I was only seven years old. Trying to understand what was happening to my little sister was very challenging for a seven-year-old, but I do remember being afraid a lot. When my grandma told me that Lola was in the hospital I remember being scared because I didn't know what was wrong with my little sister and I didn't know what was going to happen to her in the future. It took a month for Lola to be diagnosed with cystinosis.

The whole time that Lola was in the hospital was very challenging because we could only visit her and my mom a few days a week, and not seeing them a lot was hard on all of us. When Lola was diagnosed, I remember being relieved that they finally knew what was wrong with her, but it also presented me with a new set of fears about what was going to happen from there. Because cystinosis is so rare I didn't know what to expect or how it was going to affect Lola, and that feeling of not knowing what was happening was very hard to deal with. Despite everything she has been through Lola is a fun, energetic person, and she never fails to make the people around her feel good.

Lola is the bravest person I know. Even when she was struggling through hospitalization, and the numerous tests and procedures she had to go through, she never stopped being her loving self. I have always looked up to her because she has always managed to stay positive even when things were not going well, and she still manages to brighten up every room she walks into just by smiling.

A few years ago, when I was writing a speech for Lola's Curl For A Cure, I asked Lola what the worst thing about having cystinosis was. She responded, "Not being able to do some of the things that other kids get to do." My hope for Lola in the future is that she will be able to do everything she wants to do and not be held back by cystinosis. I am also very hopeful that they will find a cure so that Lola and kids like her will be able to live normal lives away from this disease.


Lola's
Curl
FOR A
Cure

Courage

"Courage is not having the strength to go on; it is going on when you don't have the strength."
Theodore Roosevelt

By Teresa Partington, Jenna and Patrick's mom
SACRAMENTO, CALIFORNIA

Jenna loves quotes. This is one that she printed out and placed in the front cover of her three-ring binder for her junior year at school. Seeing this in Jenna's backpack on the first day of school broke my heart a little. What a deep and very real thought for Jenna… who continues to persevere in spite of the hurdles placed in front of her. Patrick, who doesn't relate as much to quotes and outside inspiration, relies on his very quiet, contemplative and determined way of being to keep himself above the waterline. As Jenna and Patrick look toward being 17-year-olds, they are becoming their adult selves and it's glorious to watch. Cystinosis touches the lives of our family every day, and on some melancholy days, it dominates. Jenna and Patrick must have so many thoughts and concerns and feelings about the disease they live with, yet they press on bravely every single day. We've always said we'd choose happy over healthy, and the twins are happy young people.

I missed the window to write for the last magazine. Jenna was preparing for her second osteotomy, COVID-19 was spreading worldwide, nobody was leaving the house and it was overwhelming to think of putting all of it into words. Sometimes keeping cystinosis in the back of my mind is my way of "…going on when I don't have the strength". Like so many people in the world last year, I was trying to keep it together. I just couldn't bring myself to write it all down.

This school year is already better! A visit to Shriners last week found both kids at the likely completion of their orthopedic care. Eight surgeries (and brave recoveries) were divided between the two kids between their 6th and 10th grade academic years. This year feels much more normal as it relates to COVID-19 and cystinosis. Patrick is driving and will have his license in October (Jenna is still working on it!). He and Kevin recently enjoyed the memorable father-son experience of finding Patrick's first car. The braces are off. Jenna has a boyfriend (gasp!). Fall football games and school dances are back. Teenage banter, fashion and bizarre sleeping habits abound. All of these things are so typical and so, dare I say, "normal?" We are rejoicing in every moment. I sometimes wonder if these little moments would be lost on us, were it not for cystinosis, which makes things often different, sometimes difficult but so uniquely VIVID.

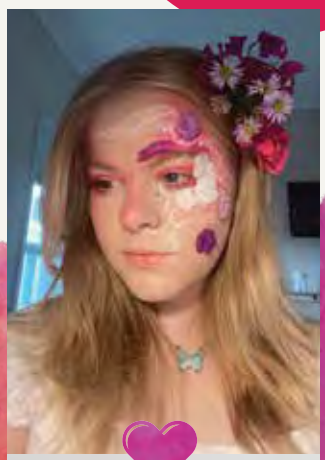
The current challenge for both kids is stamina. Inevitably, one of the two kids will end up spending a day at home during the school week, and staying on course with school is difficult. Cystinosis affects the proper functioning of

Read about Shannon Bell's Capital Cup team honoring Jenna and Patrick's Foundation of Hope on page 65.

the renal system. Patrick and Jenna both have 25% kidney function, and will receive living donor kidney transplants at Stanford University's Lucille Packard Children's Hospital in the next 12 months. Kevin is a likely donor for one of the kids (he has a few tests remaining to ensure his good health and ability to donate). The second kidney donation will come from a friend, and we are blessed to know that no less than six people are currently being vetted. It's not lost on me that for 15 years we have humbly asked our community for funding for research of cystinosis. Now, we find ourselves asking for people's organs?! It's unthinkable, yet true!

Jenna continues to love art and fashion, makeup and special effects. She looks forward to attending a Harry Styles concert in November and sings along to the music that blares from her room daily. As of this writing she very much likes a boy named Bradly, (who at the time of this writing, Kevin and I find quite charming). Jenna nurtures and communicates and loves and defends with all her heart.

Patrick is 5'9"! He swims for fitness and makes careful food choices. He takes his meds without reminding, sets his alarm for school and does his schoolwork without prompting. It's been freeing and fun to watch Patrick grow in all these ways, becoming independent and looking forward to life beyond home. Patrick is excited to take guided tours of three San Francisco colleges this week, as he ponders living in a "big city" where the weather is cool and home is less than two hours away. Jenna



CONTINUED > > >

says outright that she plans to go to college "wherever Patrick goes." While Jenna used to "mother" Patrick and call all the shots, she has come to rely on him in their more mature years. It might be co-dependent, but it's very special, their unique circumstances have given them each something that only the other can understand. To share that burden, to lean on each other and to know how each other feels - it must give them some peace. How blessed they are to have each other.

In order to be candidates for the (so far very successful!) stem cell and gene therapy treatment, the twins need to be one-year post-kidney transplant. The year 2023 could be Jenna & Patrick's first year of college and it could also be the year that they receive the groundbreaking medical treatment that we've all dreamed of for years. As we take the kids on their college tours, we keep in the back of our minds the fact that once again, Jenna and Patrick may be going about things a little differently than their peers.

Will Kevin and I be "empty nesters" in two years? There is so much to do before then. Stay tuned!

Love, Teresa, Kevin, Patrick & Jenna

The Wish Effect

2023 could be the first year of college. And it could also be the year that they receive the groundbreaking medical treatment that we've all dreamed of for years.

TOGETHER, WE ARE one

1 PURPOSE. 1 JOURNEY. 1 CURE.

COMMUNITY NEWS

The following pages celebrate the events dedicated to awareness and a cure by our cystinosis community. Together, we are stronger. Together, we are one!

In Honor of Isaac Andrews – Cottage Grove, Minnesota

PINS FOR A PURPOSE - MOONLIGHT BOWLING EVENT

By Brooke Dorma, Isaac's Aunt

On Saturday, October 2, following 19 months of waiting to gather safely together due to the pandemic, we were finally able to host the first Pins for a Purpose event in honor of my nephew, Isaac Andrews!

Team Isaac was in full force with 150 people registered at Drkula's Bowl in Inver Grove Heights, Minnesota, to support Isaac and cystinosis research. The day started with moonlight bowling, followed by a silent auction, multiple raffles and ended with a delicious spaghetti dinner. We are overwhelmed by the incredible generosity and love that surrounded Isaac. We are grateful to everyone who participated in the event and those who couldn't attend but donated to help fund a cure for cystinosis! And a very special thank you to Drkula's Bowl for their hospitality and their extremely generous contribution.

We are still working through the generous donations and cannot wait to contribute to the incredible work CRF is doing to further research and make Natalie's wish come true!

From the bottom of our hearts, we thank you for your love and generosity, *Brooke Dorma, and the Andrews Family, Dana, Ben, Isley, and Isaac.*

Internet

SUPPORT CRF BY SHOPPING ON AmazonSmile

Thank you for supporting CRF by shopping online at AmazonSmile. Because of you, CRF has received $5,584.25 from AmazonSmile as of August 2021. As the holidays draw near, our thoughts turn to sharing gifts and making memories with friends and family. Please continue to support CRF's mission to fund research for better treatments and a cure by using the SmileAmazon link below and selecting Cystinosis Research Foundation as your charity. With your help, we are giving a brighter future to those with cystinosis! *https://smile.amazon.com/ch/32-0067668*

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In Honor of Emma Grace Suetta – Etna, California

SUETTA BAKING FOR A CURE ONLINE AUCTIONS

Star Baker, Shelly Suetta, and her family decided to change things up this year, and instead of hosting their annual Lillyanna's Lemonade for a Cure Bake Sale, they created several unique online dessert auctions in honor of Emma Grace. The auctions offered incredible baked desserts to celebrate Mother's Day, local graduations, and the Fourth of July weekend. The dessert auctions turned out to be a sweet success, raising $7,336 from winning bids and cash donations!

We are grateful to Shelly, Derek, Lillyanna, and Emma for their baking skills and tireless efforts in supporting cystinosis research for a cure. Thank you, Suetta family!

Emma's Wish

In Honor of Lily Beauregard – Swansea, Massachusetts

LIVE CONCERT FUNDRAISER

Help Lily BLOSSOM

On June 27, a bright sunny, hot, and breezy summer afternoon in Rhode Island, more than 200 people gathered for the Third Annual Live Music Fundraiser to support cystinosis research in honor of Lily Beauregard. The musical performances by members of the RI Music Hall of Fame included The D'Vottes, Mark Taber and The Dick Clarks donating their time and talents to support cystinosis. We are grateful to Tom Wallis and his wife Jane, who spent countless hours organizing and coordinating the successful event. Thank you to the volunteers, musicians, vendors and the community for your contributions to help raise more than $5,657 for cystinosis research!

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In Memory of Weston Tschannen – Brookfield, Missouri

WESTON TSCHANNEN MEMORIAL GOLF TOURNAMENT

The Tschannen family and friends were excited to be able to host the 4th Annual Weston Tschannen Memorial Golf Tournament in memory of Weston Tschannen. The turnout was better this year than last year. There were many generous sponsors and golfers who together raised close to $20,000 for local organizations that Weston was passionate about, including $10,000 for CRF and cystinosis research. Thank you to the Tschannen family for your endless commitment to the cystinosis community. Together we will honor Weston's memory and find the cure!

In Honor of Landon Hartz – Pittsburgh, Pennsylvania

LOTS OF LOVE FOR LANDON CHARITY GOLF TOURNAMENT

The 9th Annual Lots of Love for Landon Charity Golf Tournament held on Friday, June 4, was a spectacular day for the 147 golfers who took advantage of the sunshine, low humidity, and cool breezes to enjoy a perfect day of golf in honor of Landon Hartz! We are grateful to the dedicated volunteers, outstanding golfers, generous donors, and crew at Blackhawk Golf Course for creating another successful golf event. The tournament raised over $26,000 to bring the total raised since 2014 to more than $158,000!

Our heartfelt thanks to the Hartz family and their team for their dedication and commitment to CRF and our mission to fund cystinosis research. You have given the cystinosis community hope – thank you!

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In Honor of Stella Grace Miller – Port Clinton, Ohio

A RAFFLE AND COMEDY FOR CYSTINOSIS!

We are thankful to the organizers of two fundraisers who helped raise money for cystinosis research in honor of Stella Grace Miller. The first event was an online auction hosted by Stella's cousin, Dylan Miller, and the PTK Honor Society at Suffolk County Community College in Selden, New York. They raffled off gym memberships and restaurant gift cards that generated $720. The second fundraiser was organized by comedian, John Butera, a friend of the Miller family, who along with his cast of outstanding comedians contributed $480. They raised money through entrance fees and raffle baskets with gift cards and adorable stuffed animals from Hershey Park. Thank you to the Miller family and their family and friends for their commitment to research. We are united in our efforts to find a cure!

In Honor of Jenna and Patrick Partington – Sacramento, California

SHANNON BELL HONORS JENNA AND PATRICK PARTINGTON AT THE CAPITAL CUP

In September, the Partington's dear friend Shannon Bell from Nor-Cal Beverage participated in Sacramento's Villara Capital Cup golf tournament for the fifth time. Each year, over 20 Sacramento CEOs gather for this tournament, with each competitor raising funds for a charity of their choosing. Shannon played for Jenna & Patrick's Foundation of Hope to raise an unprecedented $83,500 this year! The event culminates in an evening of celebration for all golfers and their charities, where Jenna & Patrick received the check with their hero, Shannon.

Thank you, Shannon, and wonderful friends in the Sacramento community who have so generously contributed more than $250,000 in honor of Jenna and Patrick and their cause to fund cystinosis research for better treatments and a cure!

Jenna & Patrick's FOUNDATION OF HOPE

In Honor of Brooke Emerson – Hammonton, New Jersey

YPTC'S 2021 MARCH MADNESS CHARITY POOL SUPPORTS HOPE FOR BROOKE FUNDRAISER

Every March, Jill Emerson's employer, Your Part-Time Controller (YPTC), holds a March Madness Charity Pool. Employees complete a bracket and if their bracket wins, the total earnings are donated to the charity of their choice. For the 2021 tournament, the YPTC employee pool totaled $1,495. The tournament was a competitive one! To Jill's surprise, Baylor University pulled through to beat Gonzaga, and her bracket won! In honor of her daughter Brooke, Jill selected the Cystinosis Research Foundation.

In addition to the pool earnings, YPTC matched the amount raised by the employees, for an overall donation to CRF of $2,990 in honor of Brooke.

Jill is so appreciative of her colleagues' and employer's support of nonprofit organizations, and specifically of her family's fight to cure cystinosis. YPTC has been behind Jill every step of the journey, from colleagues donating each year to the annual Fishing for Brooke's Cure fundraiser, to the flexibility her employer has offered her while juggling work and Brooke's disease. As an accounting firm that specializes in nonprofit organizations, it is evident that YPTC is committed to nonprofit organizations.

They are also committed to their employees as well. It truly is the best place to work! Thank you YPTC for the generous March Madness tournament donation to CRF!

Internet

CYSTINOSIS FAMILIES AND FRIENDS SUPPORT RESEARCH THROUGH FACEBOOK FUNDRAISING

We are forever grateful to all our Facebook friends who have set up fundraisers to support the important research being done to improve treatments and ultimately find a cure for cystinosis. And now, with Facebook Fundraiser, it is even easier! Just type in the link on your browser to get started. *www.facebook.com/fund/CystinosisResearchFoundation*

Since January 2021, our Facebook Community has raised $30,928 to support cystinosis research. Because Facebook doesn't charge fees on fundraisers for nonprofits, the money you raise for CRF will go directly to cystinosis research. CRF will put your contributions to work immediately when the 2021 Fall Research Grant Awards are announced at the end of the year. Together we are creating a brighter future for the cystinosis community!

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In Honor of Hadley Alexander – Boise, Idaho

"WINE NOT GIVE"

Hearts for Hadley was selected for the April "Wine Not Give" event at Split Rail Winery. In previous years the entire winery would have been filled with supporters, but due to COVID-19 restrictions, only 30 guests were able to attend the in-person event in Split Rail's Boise tasting room. Split Rail donated 30% of all wine sales at the event plus online orders. The event was an enormous success and raised more money than any previous "Wine Not Give" event. Split Rail donated $1,494 to Hearts for Hadley for cystinosis research. CRF is grateful to everyone who participated to support Hadley and cystinosis research.

BRIT & BELLE

Friends of the Alexander's who own Brit & Belle, an online accessory boutique, selected Hearts for Hadley to receive 10% of their Boise trunk show and online sales for April 1st and 2nd. The trunk show took place at the spacious home of Currie Bucher, aka the Belle. There was a bar set up outside that served prosecco and "Heartinis" so attendees could shop and sip! The event was a success and raised $600 for Hearts for Hadley and cystinosis research.

CYSTINOSIS COMMUNITY CALENDAR OF EVENTS

SAVE THE DATE TUESDAY, NOVEMBER 30, 2021

Mark your calendars for Giving Tuesday! Cystinosis is a relentless, chronic disease that affects every part of the body. CRF is the leading fund provider of cystinosis research in the world; we have singularly created a vibrant global research community. So, on November 30, let's come together to keep the research going. Every dollar you donate will support researchers who are working to find better treatments and, potentially, a future without cystinosis!

Help Fund a Cure for Cystinosis! **www.CystinosisResearch.org**

We would like to acknowledge all families for their support of cystinosis research, unfortunately some events may have passed by the time this issue is mailed.

Thursday, March 10 – Friday, March 11, 2022

CRF INTERNATIONAL CYSTINOSIS RESEARCH SYMPOSIUM *By Invitation Only*
Beckman Center, Irvine, California

Saturday, March 26, 2022

2400FT OF SCHWEITZER
24 HOURS FOR HANK, HENRY STURGIS
Schweitzer Mountain, Sandpoint, Idaho
Contact Brian Sturgis, bsturgis@simulstat.com

Thursday, March 31 – Saturday, April 2, 2022

CRF DAY OF HOPE FAMILY CONFERENCE
Balboa Bay Resort, Newport Beach, California
Contact Nancy Stack, nstack@cystinosisresearch.org

Saturday, April 2, 2022

NATALIE'S WISH CELEBRATION AND FUNDRAISER
Balboa Bay Resort, Newport Beach, California
Contact Zoe Solsby, zsolsby@cystinosisresearch.org

March or April 2022

7TH ANNUAL FISHING FOR BROOKE'S CURE
HOPE FOR BROOKE, BROOKE EMERSON
Dates and Fishing Locations to be Determined
Contact Clay Emerson, clay.emerson@gmail.com

Friday, April 29, 2022

10TH ANNUAL SWING GOLF EVENT
JENNA & PATRICK'S FOUNDATION OF HOPE
IN HONOR OF JENNA AND PATRICK PARTINGTON
Catta Verdera Country Club, Lincoln, CA
Contact Kevin Partington, kevin.partington@cushwake.com

Friday, July 22, 2022

SPORTING CLAYS SHOOT FUNDRAISER
HOPES & WISHES FOUNDATION,
IN HONOR OF JAKE KRAHE
Medina, Ohio
Contact Amy Krahe, ajkrahe@gmail.com

Monday August 15, 2022

AIDAN'S ARMY GOLF TOURNAMENT
IN HONOR OF AIDAN O'LEARY
Orchard Lake Country Club - Orchard Lake, Michigan
Contact Erin O'Leary, erinkmccarthy33@gmail.com

Thursday, October 21, 2022

SETH'S CIRCLE OF HOPE
IN HONOR OF SETH deBRUYN
Calgary, Canada
Contact Kristen Murray, murraykristen@hotmail.com

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cystinosis RESEARCH FOUNDATION

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The mission of the Cystinosis Research Foundation is to find better treatments and a cure for cystinosis by supporting bench, clinical and translational research. Since 2003, CRF has raised nearly $62 million with 100% of your donations going to support cystinosis research.

EDUCATION

The CRF is dedicated to educating the medical and public communities about cystinosis to ensure early diagnosis and proper treatment.

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