

cystinosis magazine



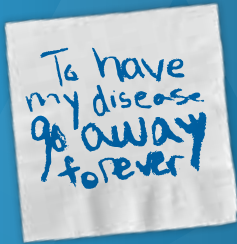
FOR FRIENDS AND SUPPORTERS OF THE CYSTINOSIS RESEARCH FOUNDATION

SPRING 2020



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 (EC cystagon, RP 103 and now 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 transplanted on October 7, 2019.

2020

- Road to a cure! Today, CRF is the largest fund provider of grants for cystinosis research in the world, issuing 190 grants in 12 countries.
- CRF has raised more than \$55 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.

DESTINATION CURE

The first stem cell and gene therapy clinical trial for cystinosis is under way! The Cystinosis Research Foundation is grateful and motivated as we navigate this road to the cure. We are actively making progress in so many ways!

Together, we are leading the way to Destination Cure.

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.

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NEW ADDRESS

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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 over \$55 million for cystinosis research in an effort to find a cure.



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SPRING 2020

DEAR FAMILY AND FRIENDS



The world as we know it has certainly changed in the last couple of months. I hope that by the time you read this magazine, we will be over the worst of the coronavirus pandemic and life will take on a more normal cadence and pace.

Like all of you, the pandemic affected the operations of the foundation and we needed to adjust. For the most part, we have worked remotely and we have become experts at Zoom meetings! We kept the research going and even received new grant applications which will be funded later this year.

We adjusted to the new normal because the survival of mankind depended on all of us working together to help each other. We have learned a lot about ourselves and our capabilities and have found hope and joy in the long days of social isolation. I believe what kept most of us going week after week was the thought of seeing our family and friends again, in person and celebrating life as a family and community.

In early March, as the threat of the coronavirus pandemic loomed, we cancelled the CRF Day of Hope family conference and Natalie's Wish event. We are glad we did although it was a

heartbreaking decision. The highlight of our year is the family conference. It is a time for cystinosis families and patients to join as one and learn about the research CRF funds, meet the clinicians and scientists who so lovingly care for our community and celebrate the progress towards the cure. We have encouraged more virtual social interaction and focused on next year's event which will be an enormous celebration of life!

WE HAD A TREMENDOUSLY SUCCESSFUL 2019 AND AS A COMMUNITY, WE HAVE SO MUCH TO BE THANKFUL FOR – OUR DONORS, OUR FAMILY AND FRIENDS, AND THE RESEARCHERS AND SCIENTISTS WHO REMAIN COMMITTED TO FINDING BETTER TREATMENTS AND A CURE FOR CYSTINOSIS.

We thank our CRF-funded researchers, scientists and clinicians whose tireless work has helped transform the understanding and treatment of cystinosis. When we started CRF, there were only a handful of researchers studying cystinosis but today, because of CRF, there are now hundreds of cystinosis researchers around the world. CRF has created a thriving and collaborative international community of researchers who have dedicated their careers to finding better treatments and a cure for cystinosis.

Natalie's birthday wish, "to have my disease go away forever" was written 17 years ago and remains our motivating force. Her wish became the wish heard around the world and has been a rallying cry for our community. CRF's aggressive research agenda has transformed Natalie's wish into clinical trials and new treatments.

COULD THIS BE THE CURE?

UPDATE ON THE STEM CELL AND GENE THERAPY

We began funding Dr. Stéphanie Cherqui at UCSD in 2007 and have issued grants totaling over \$5.56 million for her research. Since that time, the seed money we provided has been leveraged by other funding agencies including CIRM and the NIH. Although it was not an easy road to FDA approval, Stéphanie remained focused on her research and now, the clinical trial is a reality.

The Phase I and II trial of the genetically-modified autologous stem cell transplant was approved by the FDA in 2018 for six adults with cystinosis. The treatment involves taking hematopoietic stem cells from the patient and genetically modifying them with a lentivirus vector to insert a correct copy of the cystinosis gene. These stem cells (without cystinosis) are then transplanted back into the patient.

The first stem cell transplant officially occurred on October 7, 2019, when Jordan Janz, the first patient to volunteer for the clinical trial, was transplanted with his genetically repaired cells. Jordan was in the hospital for almost three weeks after the transplant and then remained in the San Diego area with his mom, Barbara Kulyk, for an additional three months before returning home to Consort, Canada in January 2020.

We have good news to report about Jordan's early results! In summary, Jordan's cystine levels (which are used to measure the amount of cystine in the white blood cells) are low which is excellent! A person with cystinosis must take cysteamine to control and lower cystine levels. *Jordan has not had cysteamine, the life-saving treatment to lower his cystine levels, since September 2019! At this point, six months post-transplant, his cystine measurements are low enough that he does not need to take cysteamine therapy. The genetically repaired cells are doing their job!*

In Jordan Janz's own words:

"Being where I am today couldn't have been accomplished without so many things falling into place like Stéphanie Cherqui coming up with the stem cell transplant and Nancy creating Natalie's Wish. Also, every single person out there who has donated their time and/or money to help make this possible. And without the amazing cystinosis families and the things they have done, I wouldn't be sitting here today not having to take 58 pills a day! Right now, I am taking 18 pills a day, which is 40 less than before I had the stem cell transplant and I am not taking cysteamine! So, for that, I thank everyone and pray that every person will get cured and cystinosis will be a thing of the past."

A SMALL SETBACK FOR THE STEM CELL TRIAL

As the health care community responded to the anticipated increase in coronavirus cases, UC San Diego Health decided to postpone all elective procedures and clinical trials. While we regret this recent development, *a second patient in the study has been enrolled and has completed apheresis.* Cryopreserved drug product for that patient has been manufactured and dosing is expected to occur as soon as the UCSD clinical site allows. We are optimistic that we will be able to resume the trial once the coronavirus response has been settled. We will keep you updated on all ongoing developments.

We thank Jordan for volunteering to be the first patient to undergo the treatment and we thank patient #2 who is ready to be transplanted once UCSD resumes clinical trials. Thank you to our family and friends who believed that this day would come. We hope and pray that the positive trend that Jordan is experiencing continues and that this one-time stem cell treatment will stop the progression of cystinosis or be the cure!

OTHER RESEARCH UPDATES

CORNEAL CYSTINOSIS

Corneal cystinosis is the build-up of cystine crystals in the eyes that causes photophobia, severe eye pain, and sometimes, blindness. There is an existing eye drop treatment but the drops must be taken every waking hour and for many patients, the drops are painful to administer.

For the past few years, we worked on a potential treatment for corneal cystinosis using a nanowafer as the delivery system for cysteamine, the medication to treat corneal cystinosis. After years of work and funding, we had to cut our losses and end the program. The technology was not replicable and although we tried to work around the issues, we realized that it would be impossible to do. We are very disappointed with this outcome; however, we had a talented team dedicated to the project and we did everything in our power to find a solution.

SOLIDROPS - MORGAN FEDORCHAK, PhD

We continue to be impressed with the work of Morgan Fedorchak, PhD, who is at the University of Pittsburgh. Dr. Fedorchak has developed a remarkable delivery system for cysteamine as a treatment for corneal cystinosis. The SoliDrop, is a thermoresponsive hydrogel that contains spray-dried, cysteamine-loaded microspheres that turns from a liquid to a gel once it is dropped into the eye. We believe that one drop per day with a controlled release formulation of cysteamine will provide a full

day of therapy. Please read Dr. Fedorchak's informative question and answer interview on page 42.

CRF has committed to funding Morgan's work throughout the next critical phases which include final formulation, animal studies and the IND (Investigational New Drug) application. We plan to exercise our Option Agreement with the University of Pittsburgh to acquire an exclusive license to the technology for use in developing a new FDA approved treatment for corneal cystinosis.

A UNITED COMMUNITY OF SCIENTISTS AND RESEARCHERS

CRF was proud to sponsor the seventh International Cystinosis Research Symposium on February 26-28 in Irvine, CA. A sincere thank you to the co-chairs of the symposium, Corinne Antignac, MD, PhD, Julie Ingelfinger, MD and Stéphanie Cherqui, PhD who organized and planned the symposium. CRF-funded researchers and scientists from seven countries attended the symposium to present their research study updates and progress. We heard exciting updates about potential new therapies for Fanconi syndrome, muscle wasting and ocular cystinosis. Several scientists shared new models for studying the disease, including genetically engineered rats, zebrafish and stemcell-derived organoids. It was a very productive and stimulating conference that yielded new ideas about cystinosis and several research collaborations. We are grateful to all CRF researchers for their dedication to finding better treatments and a cure for those with cystinosis. Please read more about the symposium on page 57.

TEN NEW GRANTS TOTALING \$2,617,711 ISSUED TO-DATE

CRF funds new grants twice a year to ensure that your donations are always funding research.

We are pleased to announce that in 2019, CRF issued ten new grants totaling \$2,617,711 million for cystinosis research. The grants were awarded to researchers in the United States, Belgium, France and Italy. In the fall of 2019, we issued three new grants for a total of \$856,488. The three new recipients of the 2019 grant awards are listed on page 50 along with a lay abstract of their study.

Since 2003 CRF has funded 190 multi-year research studies in 12 countries. Our researchers have published 82 articles in prestigious journals as a result of CRF funding. CRF is the largest private fund provider of cystinosis research in the world. Our grants have resulted in several clinical trials.

Given the breadth of research currently funded, you can be assured that there will be more breakthroughs and life-changing treatments and discoveries in the near future. You have been with us every step of the way and we are forever grateful. We could not do this without you.

CRF RESEARCH HELPS OTHER DISEASES AND DISORDERS

Your support of cystinosis research reaches far beyond our small community of cystinosis. Our discoveries are being applied to other more prevalent diseases and disorders. Discoveries made by our researchers have the potential to help millions of others with diseases and disorders similar to cystinosis. In fact, discoveries from our research teams are being applied to Friedreich's Ataxia, Danon disease, corneal diseases, kidney diseases and genetic and systemic diseases similar to cystinosis.

GRATITUDE AND HOPE

I was honored to be in the room when Jordan was transplanted. It is a day I will never forget as I watched the two small clear bags of repaired cells be transfused into Jordan. All of the years I have spent living with cystinosis raced through my mind. I thought of the day our daughter Natalie, made her wish and how on October 7, 2019, her wish became a reality because of the heroism and bravery of Jordan Janz. How blessed I felt to be in the room that day sharing that life-changing moment with Jordan and his family and Stéphanie Cherqui. Only time will tell if the transplant is a cure but the early results are promising.

As the world suffers through the pandemic, I am sure many of you have survived the disease and some of you have lost people you know and love to COVID-19. We have been united in our suffering and we have searched to find meaning and hope in the challenges that face us. Together we have faced the challenges of cystinosis, we have overcome obstacles and sometimes we have had to alter our strategy to find the best path forward. We have done this with you by our side. You have never wavered; you have supported us throughout the years. You believed in us and we are grateful for your partnership.

Thank you for embracing our community, for supporting our research efforts and for always believing that we could make Natalie's wish come true. You are part of our family, and we are fortunate to have you by our side.

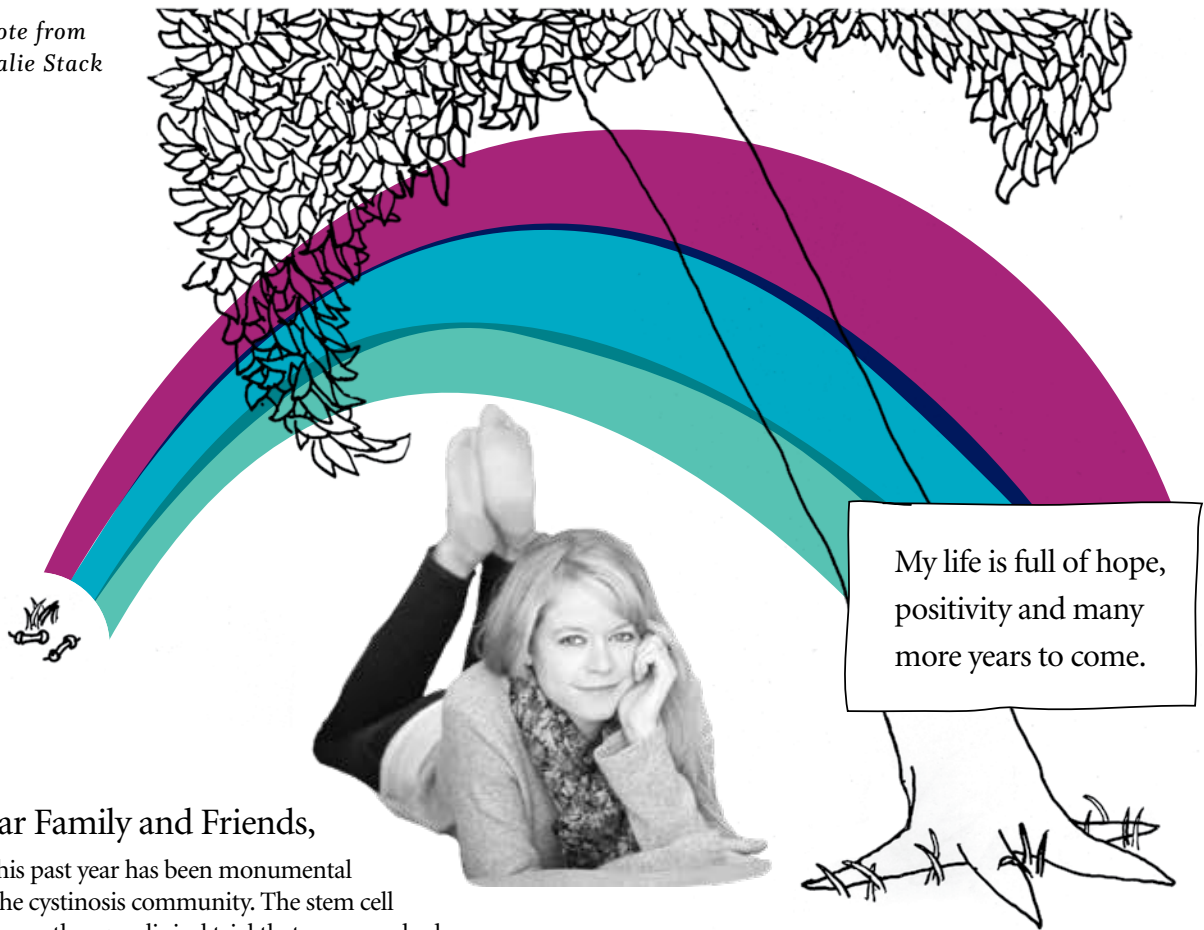
We have made extraordinary progress and with your continued support, generosity and love, we will cure cystinosis.

With heartfelt thanks and gratitude,

Nancy & Jeff



A Note from
Natalie Stack



Dear Family and Friends,

This past year has been monumental for the cystinosis community. The stem cell and gene therapy clinical trial that everyone had anticipated is finally recruiting more patients, and the first patient is doing extremely well so far. The progress and success of the stem cell trial has made me even more hopeful about my future than ever before - it is a true miracle.

Life is looking more and more positive for me each day. My overall health is great — I have not had a kidney transplant yet and am hoping to be in the clinical trial someday in the near future. I still live in Irvine with my cat and am close to my family. I have been a case supervisor at CASA OC for over a year now and I have been able to challenge myself in my position. I finally found a job that connects me with my passion to advocate for children in the foster care system. My life is full of hope, positivity and many more years to come.

Cystinosis is a horrible disease. It is unimaginable what the patients and their families have to go through every day — around the clock medications, eye drops every hour, uncomfortable stomach pain, extreme fatigue, g-tubes, rickets, sensitivity to light and the list goes on. For me, the side effects of the medication and the crystals in the eyes are unbearable at times but, even with so much pain, our community has never given up hope on a better future for us. Being surrounded by an incredible community has given me the hope and faith that cystinosis will not be my forever, and now, I believe that my future will soon be a life without cystinosis.

One day soon, I will no longer have to worry about my foul smell, my extreme fatigue, the timing of medications, my muscle weakness, my eyes hurting, my excessive thirst, my annual doctors' appointments, and most importantly, I won't have to worry about how long I have to live.

We have been so fortunate to have Stéphanie Cherqui, PhD dedicated to our community and to finding a cure. I am thankful for her commitment to the research and for her tireless effort that has resulted in FDA approval of the stem cell trial. I take comfort knowing that cystinosis will never prevent me from reaching my goals and aspirations in life. I know that I will live longer now, and, because of that, I am extremely grateful for what Dr. Cherqui, my mom and the rest of this community has done for not only me, but for all the other patients with cystinosis.

Words cannot express how blessed I feel to know each of you. Thank you for never giving up on my wish to have my disease go away forever.

Love,
Natalie

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 500 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 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 190 grants in 12 countries.

CRF has raised nearly \$55 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.



Thank You!

You have changed the course of cystinosis!



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

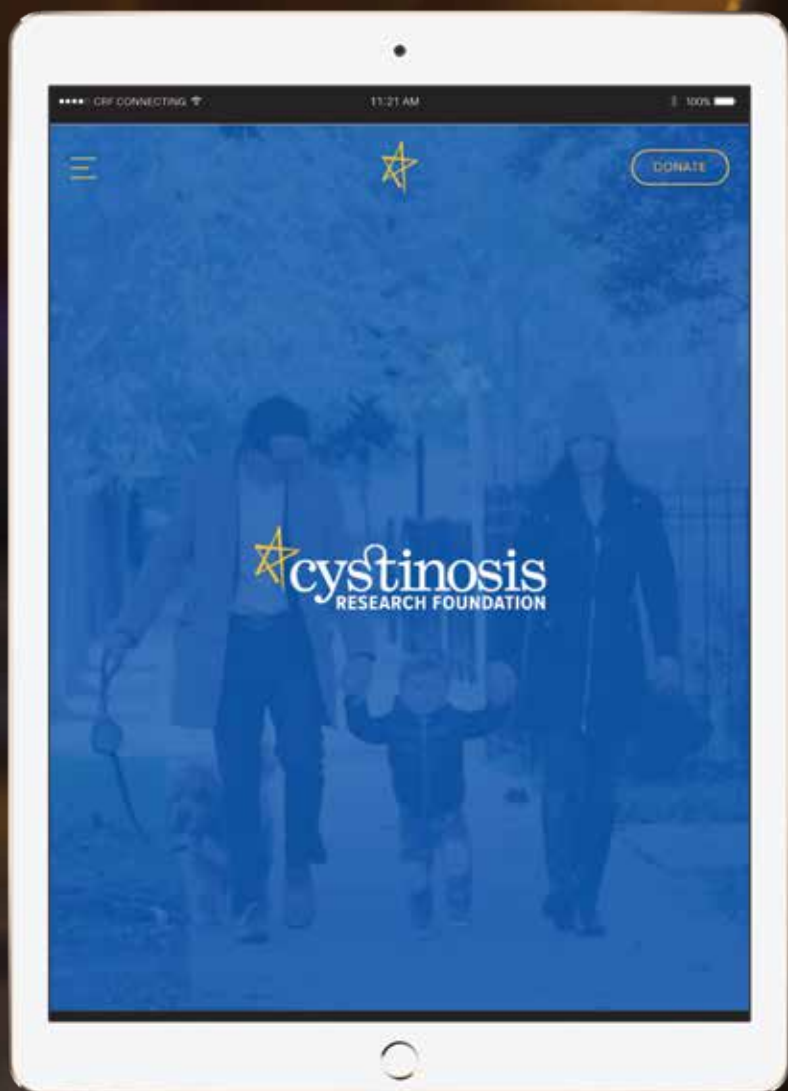
CRF's highly strategic approach to funding has resulted in two FDA approvals and several human clinical trials. The research dollars we have invested have been leveraged by over \$24.2 million in grants from other funding agencies. Not only does CRF research help our community, but our discoveries are 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.

WE'RE SHINING BRIGHT ONLINE!

We are excited to share our newly redesigned Cystinosis Research Foundation website! The site has a brand-new look and is easy to navigate. Learn more about the research CRF funds, the families who are part of our community and the events that support our research program. If you are a researcher, you will find everything you need to know to apply for a grant. Explore the site, view our collection of videos, read the archive of published papers and learn more about what we do every day to find the cure.

Visit us today, www.cystinosisresearch.org and share with your cystinosis community!





Cure Cystinosis International Registry

The Cystinosis Research Foundation is excited to announce a new Cure Cystinosis International Registry (CCIR) will be launching soon. This improved patient-based international registry will be a central hub of information created by those living with cystinosis, their families and their caregivers. The deidentified information provided by patients and their families will be shared with cystinosis clinicians, researchers and scientists who are pursuing better treatments and a cure for cystinosis.

For patients, CCIR will provide an opportunity for involvement in research that will help develop and test new therapies and develop a cure for cystinosis. 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 in an effort to accelerate better patient care.

Watch for upcoming announcements to register and be part of the cure!





BUS STOP

JORDAN JANZ'S JOURNEY

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16 BARBARA KULYK



CLINICAL TRIAL IN PROGRESS

FOREVER *Intertwined*

by Dennis Arp

Those who lived it
bring insight into
the moment when
Dr. Cherqui's gene
therapy realized
its full potential to
transform lives.

Pushed by the steady hand of a specially trained nurse, the gene-corrected cells of Jordan Janz flow through a syringe and back into his body with measured, methodical ease. As Dr. Stéphanie Cherqui watches at Janz's bedside, thoughts and memories flood her mind at a much faster pace.

Dr. Cherqui pioneered the gene therapy that in this very moment is moving from the lab to human trial. And an otherwise simple hospital room overflows with a cascade of promise.

"I thought about Jordan and his family, but I also thought about all the research, all the people, all the work over all the years," recalls Dr. Cherqui, associate professor of pediatrics at the University of California, San Diego School of Medicine. Dr. Cherqui was there to support Janz in his cell-transplant moment along with Cystinosis Research Foundation (CRF) President Nancy Stack as well as Jordan's mom, Barb Kulyk, and Jordan's dad and stepdad.



CONT'D >>>



FOREVER *Intertwined*

“Going from bench to bedside, all the research goes to the patient, it’s almost like a birth, a delivery,” Dr. Cherqui adds. “It’s this special moment you work toward for so long. I looked at Nancy because we have shared so much on this journey.”

Stack founded CRF in 2003, and the foundation first started funding Dr. Cherqui’s research 13 years ago. At that time, it

was just an unproven pipedream, this idea



It was a sacred moment for me. Our work had come full circle.

that by genetically modifying a patient’s stem cells to correct the cystinosis gene and then reinfusing them, it could effectively serve as a cure for the disease.

But ever since the project began, such possibilities have been an enduring source of hope for the entire cystinosis community.

“It was an honor to be asked by Barb and Jordan to be there for the transplant,” Stack says. “When the moment came to start pushing the stem cells through the IV it was

so quiet in the room. I remember wondering if Jordan would ever have cystinosis again. I tried to wrap my mind around how this could be the start of a treatment that could literally save our children. It was a sacred moment for me. Our work had come full circle.”

In the months since Janz’s transplant in October 2019, he has faced physical challenges as he endured the chemotherapy that preceded the transplant. More recently, an appendectomy that was unrelated to the

procedure landed Janz back in the hospital, this time in his native Canada. But now he has returned to work in his hometown of Consort, Alberta, and the prognosis is trending exceedingly positive.

“What we’ve seen in the three months after the transplant is a dramatic reduction in Jordan’s average granulocyte cystine level – a decrease of more than 80 percent compared to baseline,” Dr. Cherqui says. “That drop is what we were hoping for, but we didn’t know if we could reach it in humans.”

The day-to-day difference in Janz’s life is also significant. He has gone from a regimen of 58 pills a day to as few as 16. The biggest reason for that dramatic drop is that he has

CLINICAL TRIAL IN PROGRESS



been able to stop taking cysteamine, which means he no longer has to deal with a litany of side effects.

“After seeing the results, we know that something is happening, but we still need time to evaluate and understand it,” Dr. Cherqui says of the transplant. “I’m always cautious about everything – I’m always thinking, ‘Let’s wait and see.’ But there’s no doubt that it’s exciting to already be seeing successful outcomes three months after transplantation.”

As the phase I/II clinical trial moves forward, Janz will return to San Diego periodically for testing to assess his progress. That’s on top of regular blood draws done near his home in Canada.

Meanwhile, Dr. Cherqui’s gene therapy project is moving toward a transplant involving a second patient. No date has

been set for the second procedure, but the trial’s success so far portends a steady flow of new patients and new transplants.

“We are learning so much from the first experience,” Dr. Cherqui says. “There is an inevitable learning curve with the first patient. We’re so thankful to Jordan for his bravery and maturity throughout the process. He’s an amazingly strong young man who has been thinking the whole time about how what he’s doing will affect the whole cystinosis community.”

Janz, 21, is heartened by the prospect that this one-time therapy may eliminate the need for a kidney transplant as well as other complications related to cystine

accumulation in the organs of cystinosis patients.

“But he’s scared to say that the transplant worked,” Kulyk says of her son. “It’s hard to think about going

back to the way things were.”

Still, Janz says he’s starting to let himself consider the possibility that the treatment is a cure.

“I think about that every day,” he says. “I haven’t taken (cysteamine) pills in six months. This could be my life now. This could be what it means to live a normal life.”

*This could be
my life now*

CONT'D >>>



CLINICAL TRIAL IN PROGRESS

Barbara Kulyk

But even if it turns out there are more setbacks along the way, Janz is glad that he assumed the responsibility of being patient number one for the trial.

“I certainly want to share my experience and help others make the decision that’s best for them,” he says.

Before he committed to the trial, Janz first had to get past the uncertainty. He needed time and information to find his way to a yes. The procedure is a lot like

a bone marrow transplant but in this case, the patient is both donor and recipient, so there’s a low risk that the body will reject the re-engineered cells. And at a time when Janz was feeling relatively healthy, he had to prepare himself to endure the effects of the procedure.

“The decision took me about a month, once I went through the forms, read every day about the possible side effects and weighed the negatives against the positives,” said Janz, who was initially diagnosed with cystinosis as an infant.

There were moments in which he thought, “Why put my body through this when there are no guarantees,” he admits. “At the same time, I thought about how someone has to

do this. By reading up on Stéphanie and her system, I became confident that it was safe. I had to try it.”

The hardest part was the chemotherapy, which was needed to open space in his bone marrow so his body could accept

the transplanted cells. In addition to nausea and hair loss, Janz endured painful sores inside his mouth, preventing him from eating and speaking normally for the 10 days when the effects were at their worst.

“It was especially hard on Jordan because he loves to eat,” Dr. Cherqui says. “But even on his worst days, he still wouldn’t complain. He was still trying to talk to me when I would come in to see him.”

“I had no idea he was this brave,” adds Kulyk of her son.

Throughout the transplant process, Kulyk has been at Jordan’s side to support him. When she first heard of the trial, she made a call that got Jordan considered and ultimately identified as an ideal candidate. She has constructed a support system for her two younger children and made sacrifices in her career so she could join her son for all the trips from Alberta to San Diego. She has read the research

*It's a moment
we've chased
for 20 years*



Dr. Midgley and Jordan Janz



Jordan Janz and Jeff Kulyk

FOREVER *Intertwined*

and answered his questions -- given him counsel and comfort when each was needed most.

Now that she sees him thriving, she's also allowing herself to harbor real hopes that her other children won't have to be asked to donate a kidney.

"I don't know why it has happened for us, but we're humbled and grateful that Jordan was chosen," Kulyk says. "It's a moment we've chased for 20 years, and now that we've caught it, it's a weird feeling. When we first got home (to Alberta), I kept checking things because I had a weird feeling, like I had left the tub running or the garage door open."

Kulyk further describes a "feeling of one" -- something like what she experienced when Janz was first diagnosed and she didn't yet know any other cystinosis families. Except that now the oneness comes with a desire to share feelings of hope.

"I try to step up to the plate when other parents call me, just like Nancy did for

me when I first called her," Kulyk says. "I try to lead by example."

For Stack, this is a moment of abundant gratitude for the countless heroes who have stepped up over the years to make progress possible.

"I keep thinking about what a brave, selfless and courageous young man Jordan is to volunteer to be the first person to have the transplant," she says. "How can I thank him? There are no words."

Like Dr. Cherqui, Stack is recognizing extra resonance in experiences that rise from the mundane to the historic, given the momentum of the moment. Watching amid the silence as gene-corrected cells re-enter Janz's body is just such an experience.

"I thought about the word hope and what that meant for all of us in the cystinosis community," Stack recalls as she puts herself back in the transplant moment. "Hope has driven our research. We raised the money and then we needed the talent, and we found it by

There is nothing more powerful and fierce than the love a parent has for their child.

CONT'D >>>

CLINICAL TRIAL IN PROGRESS



FOREVER *Intertwined*

funding one study at a time. Our community of donors has been steadfast in their support of our research efforts. Our CRF Scientific Review Board kept us on track and recommended only the best research projects for us to fund.”

Stack’s thoughts continue to pour forth.

“I thought about Stéphanie and meeting her in 2006 and hearing her say that she would work until she found a cure for our children,” Stack says. “I thought about how fortunate the cystinosis community is to have Stéphanie in our lives, battling for our children every day. She should win the Nobel Prize.

“I thought about all of the cystinosis families who raised money for research or helped by supporting CRF. People had big fundraisers and small fundraisers; everyone wanted to be part of the cure. There is nothing more powerful and fierce than the love a parent has for their child.

“I thought about my own daughter Natalie, who in 2003 so innocently made the wish ‘to have my disease go away forever,’ and how that little girl’s wish turned into an urgent movement to find a cure for cystinosis. I think about cystinosis every day and worry about Natalie’s health and future every day. On the day of the transplant, however, the worry was lighter, and the future seemed brighter. Jordan was making Natalie’s wish come true, and the wish of all of the adults and children with cystinosis come true.”

Sharing in the day of the transplant was nothing short of transformative, Stack relates.

“October 7, 2019 will forever be etched in my memory, and I will always feel blessed to have been part of it,” she says. “It was incredibly emotional, and as I told Barb the next day, ‘I am bonded to you and Jordan for life, we are family.’ We shared such an intimate, profound, life-changing experience that our lives are forever intertwined.”



Jordan and his siblings

“WHEN I READ THIS YOUNG MAN’S WORDS I WAS PROFOUNDLY MOVED. HE IS WISE BEYOND HIS YEARS AND IS DOING MORE THAN WE EVER COULD TO HELP.”

INSPIRED BY JORDAN’S STORY, CRF RECEIVES ASTOUNDING \$1 MILLION DONATION.

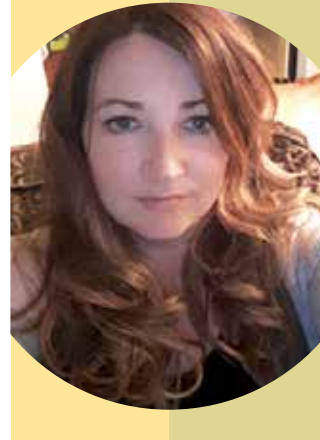
Paulette Simpson, grandmother to almost-two-year-old Charlie Simpson, who was recently diagnosed with cystinosis, read an update about Jordan Janz, the first cystinosis patient to volunteer for the stem cell and gene therapy clinical trial at UC San Diego. In the article, Jordan was discussing his role in the trial and stated, “I’m willing to do it if it helps the kids. Somebody has to do it. I don’t have the money to donate to scientific conferences and stuff like that, but I can do this trial.” After reading Jordan’s comments, Paulette was motivated by Jordan’s selfless perspective. Simpson said, “When I read this young man’s words I was profoundly moved. He is wise beyond his years and is doing more than we ever could to help. In gratitude to Jordan and his family, our family is honored to make a contribution to CRF.”

The Mary Noël Magistro Trust and Paulette Simpson donated \$1 million to CRF in honor of their great-grandson and grandson, Charlie. This remarkable and extraordinary gift will be used to continue and accelerate CRF-funded research. We are grateful for their commitment to our research program and their dedication to all children and adults with cystinosis.



WELCOME NEW BOARD MEMBER

BARBARA KULYK



My husband Jeff and I have been married for 15 years and we reside in Consort, Alberta, Canada. We have five children, Brittany (Brody), Nathan (Megan), Jordan, Victoria, Easton and two grandchildren Bridgette and Burkley. My adult children also live in Consort so I am very fortunate to have all my family around me. We are a close, supportive, loud family that likes to get together around the dinner table and discuss or argue over many topics.

In my lifetime, I have always chosen employment that worked around a chronically ill child that had a lot of needs and a strict schedule. As many of you know this can be tricky to balance. I participated in night and evening courses to earn my aesthetician certificate and upon completion I opened a spa, thus allowing me to set my own hours. When Jordan was ten years old, he was accepted into the RP-103 trials that were held in Chicago. At this time, I sold my salon and began teaching aesthetics in between trips.

Jordan turned seventeen years old around the time the trial was completed and he had become very independent in his day to day needs. By this time, I had taken a job with more responsibilities and I was employed as Executive Director for Adult Learning and Family and Community Support Services. Currently, I am the Chief Administrative Officer for Consort and have been employed for the Village for the past two years.

Many of you know that Jordan was the first patient transplanted with the stem cell and gene therapy treatment that was held in San Diego in 2019/2020. Going through this experience with Jordan and my family really put life into perspective for me. It suddenly became very clear where and with whom I wanted to spend my time. I cherish the little things and try to never take my family and friends for granted.

I am very happy about being accepted to the Cystinosis Research Foundation Board of Trustees. In the past, I haven't had much time available to reach out to others as I was trying to raise my family and meet the needs of Jordan who has (had?) cystinosis. Now that my family is growing up, I finally feel I can commit to this community on a broader level.

Being part of the cystinosis community means having the opportunity to see the beautiful little faces of the children when I attend the Day of Hope. They are all adorable and remind me so much of Jordan when he was younger. Watching them run around, I can't help but love them all. I also look so forward to seeing the adults and catching up on their lives. I feel connected to the parents without saying a word and appreciate just being in this atmosphere for a few days; I personally feel I "belong."

My hope in joining the board is that I will have the opportunity to make others affected by cystinosis feel like they are part of this family in whatever form that may take.



"THIS is what HOPE looks like." BARBARA KULYK

CRF VIDEO PREMIERE

We're Turning Positivity Into *Progress*

TOGETHER

As we live with the fear and frustration of our shared global health crises, we can reflect and appreciate the power of one little wish Natalie made so many years ago.

Even though we can't meet in person this year, that doesn't mean the CRF community can't share in the incredible story of the clinical trial that unfolded this past year.

Please join us in the comfort of your own home for the CRF 2020 Video Premiere.

www.cystinosisresearch.org/videos



A 2020 CYSTINOSIS RESEARCH FOUNDATION FILM
CRF PRESENTS TOGETHER WE SHINE BRIGHT STARRING RESEARCH HOPE CURE
CUSTOM LAYOUT BY IDEA HALL CINEMATOGRAPHY BY LARS WANBERG PRODUCED BY CYSTINOSIS RESEARCH FOUNDATION

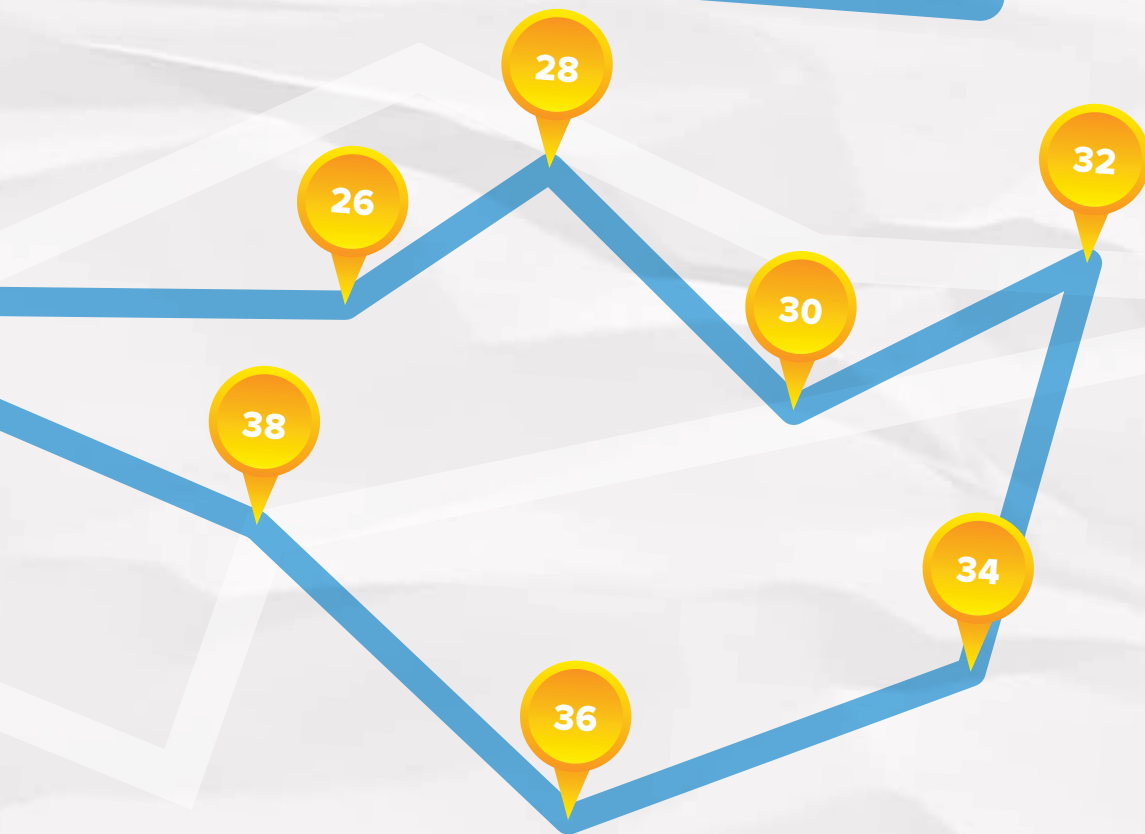


24

- 24** TINA FLERCHINGER
- 26** KURT GILLENBERG
- 28** ALEX WEAVER
- 30** HADLEY ALEXANDER
- 32** BROOKE EMERSON
- 34** HENRY STURGIS
- 36** OLIVIA LITTLE
- 38** JENNA & PATRICK PARTINGTON

FAMILY STORIES SPRING 2020

OUR JOURNEY TO A CURE





Sweet 16 Success

By Denise Flerchinger, Tina's Mom
CLARKSTON, WASHINGTON



Last year at this time, we noticed Tina's health was declining with daily migraines, lack of appetite, stomachaches and extreme exhaustion—leading her to nap six hours a day.

Tina was consuming 83 pills a day with two gallons of water barely maintaining her health. One Sunday, she invited her friends to go boating only to find she did not have the energy to get in the water or carry on a conversation. On other summer nights she would go out only to return home sick and frustrated. She was fading away before our eyes.

A call from the pediatric nephrologist informed us that Tina was in the final stage of kidney failure. The next eight months were filled with doctor visits, tests, procedures and a plethora of blood draws—which included hundreds of test tubes.

Fast forward, October 2019, Tina received the gift of life, a new kidney from her father, one week after her 16th birthday. He was a perfect match!



This part of the journey has been emotionally exhausting, bringing back memories of Tina's diagnosis. Today we are still trying to find our new normal—balancing a new set of medications and struggling a bit to boost Tina's immune system, but I am hopeful given how far we've come.

Being a teenager is hard enough without the multitude of complications that come with having a rare disease, and some days can seem unbearable, but we have the hope the Cystinosis Research Foundation has given us. And Tina's renewed energy has allowed for her to "finally live life" (her words, not mine)—going snow skiing as soon as she got the green light from her doctors, earning straight A's and fulfilling her dream to participate in a high school team sport... she's loving track and field!

Tina has overcome many obstacles with her strong faith, courage and determination. She is happy, she is thriving...and we are ready for the next chapter (stem cell treatment)!

LIFE'S WORTH LIVING JUST REMEMBER TO *Live it*



Some believe that events in individuals' lives happen for a reason. People use this statement to justify what has happened to a person; they believe that in some small way it will make that individual feel better about their situation. But when you are dealt a terrible hand, it's hard to see a future where anything remotely positive could come from it.

I was diagnosed with cystinosis at 18 months old. Growing up, my parents worked hard to make sure I was never defined by my disease. They wanted me to be seen for who I am on the inside not a sick kid on the outside. My parents made sure I took my meds on time each day, went to every doctor's appointment, encouraged me to live life, and educated me about cystinosis. I didn't need a kidney transplant until I was almost 18 years old.

Last year, on January 29th, 2019, I received my second transplant. Prior to that, I had been on dialysis for almost four years, spending half the time on hemodialysis and the other half on peritoneal dialysis (PD). For two and half years, three days a week, I would wake up at 4 a.m., go to the dialysis center, get stabbed by two giant needles, and sit in a chair for four hours being as still as possible.

There were many side effects to hemodialysis and being in an environment surrounded by people who were decades older than me was damaging to my mental health. Especially when every member of the staff was asking me, "Why are you here?" or telling me, "You're way too young to be here!" Those statements were not the best thing to hear on a regular basis. I eventually made the decision to switch to PD and even though the treatment was every day, it was still easier for me since I could do the therapy at home.

During that first year, I was depressed and just plain angry all the time. I blamed God, the universe, the doctors, pretty much anyone who I thought could be the reason my kidney failed. I shut myself off from my family, friends, everyone. It was the first time I truly felt alone. I felt like I had no one to talk to that could relate to the pain I was experiencing. I blamed myself for having lost the gift my mother risked her life to give me.

There is nothing worse than when a situation happens that is utterly out of your control. I do not believe things happen for a reason. Tragic situations happen to a person because most of the time life isn't fair. While living with a chronic illness, it can be hard and sometimes impossible, to believe you can make a negative into a positive. For almost four years I had to hit the pause button. I was seeing my friends move on with their lives, starting their careers, getting married, moving into their own place, becoming adults, while I just felt stuck. I almost felt like I was in a hallway of doors and each door led me right back to where I started. I missed birthdays, vacations, visiting friends and missed my sister's graduation from college.

If I wanted my life back, **I WAS GOING TO HAVE TO WORK FOR IT.**

After almost three years of treatment, I realized I could not just sit here and wait for good things to happen, wait for life to begin again. If I wanted my life back, I was going to have to work for it. It was hard and looking back I honestly don't know how I found the strength to do it with everything I was going through. I finished my AA degree in Social and Behavioral Sciences and I became a nationally certified clinical and administrative medical assistant during the last couple years of dialysis. It took me a long time to realize my purpose in life and to come to a better understanding of what I could give back to those in similar circumstances.

Even if I can help one person realize they have more to offer the world than just being that sick kid everyone feels bad for, then the experiences I have been through will be worth it. Cystinosis can be a debilitating disease if you let it but that doesn't mean you can't live your life to the best of your ability. This means saying "yes" to opportunities that are outside of your comfort zone. Even if you feel that you don't have the strength to do the things you want to do or if your doctors say you can't, remember it's your life.

Sometimes you need to grab the bull by the horns and hold on. Though you might feel your grip loosening, just remember, if you want a life you are going to have to fight for it. It may seem impossible at times, but once you start to live again it will be worth all the pain and suffering. I promise.



...say "yes" to opportunities that are outside of your comfort zone...

By Kurt Gillenberg

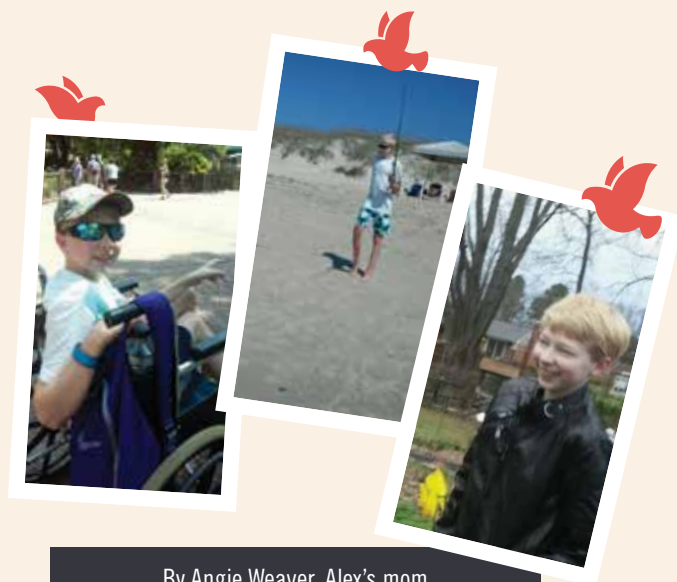
SANTA CLARITA, CALIFORNIA



*the
beginning
of many
blessings!*



Blessing after Blessing after Blessing!



By Angie Weaver, Alex's mom
MECHANICSVILLE, VIRGINIA

Alex's story is similar to most. When he was 9 months old, he stopped gaining weight. We attributed this to the projectile vomiting of almost everything he was eating. Unbeknownst to us, there was a more complex reason. After a visit with his nurse practitioner, who found that his glucose was abnormally high, we were sent to a local hospital for further investigation. In less than a week, Alex was diagnosed with cystinosis. Initially, it was a time of despair, but it was also the beginning of many blessings!

Alex began taking Cystagon® at 12 months of age. His early diagnosis was our first blessing. We had to mix it with water and put it in a syringe. I remember how awful it was getting him to swallow it. Sometimes he would throw it back up, and we would do it over again. He quickly learned to swallow his pills! Another blessing!

Blessing!

Once the reality set in, I started doing my own research. I called different experts throughout the United States, including doctors, support groups and other parents of children with cystinosis. The support was tremendous. Blessing! I also called various universities and spoke with researchers who were studying gene therapy. I was told that gene therapy would not work for cystinosis. This was in 2004. I felt so discouraged. As a parent, I wanted to be able to fix this. My discouragement gave me the drive to go back to school to become a nurse. I now realize this was my own therapy. It somehow gave me a feeling of control. Those days were tiring, but we got through.

Blessing!

Alex just turned 17 this past November. Blessing! The main challenge he has dealt with has been with his bones. He had eight plates to correct severe knock knees when he was

about 8 years old. After that surgery, he was diagnosed with scoliosis. He wore a brace for many years but ended up having to undergo a spinal fusion last February. As I write this, he is recovering from hip surgery to fix his left femoral neck fracture. He fractured his right femoral neck a little over a year ago. So now he has screws in both hips! Crazy thing is, we have no idea how either of the fractures happened, there were no falls or trauma. Alex has tolerated all of his surgeries amazingly well!

Blessing!

Our journey has been full of optimism knowing that we have the Cystinosis Research Foundation on our side. The amazing families, researchers and donors have provided us with enormous hope, bringing the search for a cure to new and uncharted territories. We realize we are so BLESSED to be a part of this! Thank you for all that you do!

Cheers to Raising Money for Research

By Marcu Alexander, Hadley's mom
BOISE, IDAHO

Most Americans associate Idaho with potatoes. That may be true, and potatoes are delicious, but we're known for so much more than just the spud. Idaho, particularly Boise, is also known for having amazing local wineries and craft breweries. Our family is fortunate to be friends with several local winery and brewery owners who are caring and generous.

Over the past few months, Hearts for Hadley has held three separate fundraising events at local wineries and a brewery for cystinosis research.

The first event was a glassybaby pop-up shop held at Potter Wines on November 2nd. Potter Wines is owned by the parents of one of Hadley's best friends. Hearts for Hadley received a portion of sales from both glassybaby and Potter Wines during the three-hour event. Everyone had a great time enjoying a glass of wine or a wine tasting while shopping for beautiful glassybaby hand-blown candle holders. We're grateful to the Potter's for hosting the event.



Our next event, Kegs 4 Kause at Payette Brewing Co. owned by our friends, Mike & Paige Francis, took place on November 25th. Every Monday throughout the year, Payette Brewing Co. supports a different local non-profit through their Kegs 4 Kause program by donating 50% of beer sales between 6pm-10pm. This year we raised \$680 for cystinosis research. Payette Brewing Co. has supported Hearts for Hadley for years and we're incredibly thankful for their ongoing support through Kegs 4 Kause and beer donation for our annual Hearts for Hadley gala.

Our latest fundraising event took place on January 15th at Split Rail Winery, owned by our friends, Jed & Laura Glavin.

Every month Split Rail selects a local non-profit for their Wine Not Give program where 30% of wine sales are donated. In addition, they open their awesome tasting room for the event and let the non-profit decide how they want to raise money and awareness. We hired a food truck for our event and hired a couple of bands to entertain our guests. The opening band, Cheaper Sneakers,



was a highlight. The band is comprised of Hadley's older sister Stella, my nephew, Porter and our good friends, Henry and Nika. They all attend Boise Rock School and were excited to play at a real gig! They did a great job and were paid in grilled cheese sandwiches from the delicious food truck, Melt. The main act was a band call Tower 9. My brother-in-law plays guitar for Tower 9 and the band is truly great! The entire tasting room was packed, and the local news even showed up to film a piece about Hadley and cystinosis. We raised \$735 at our Wine Not Give event which was a record for Split Rail Winery!

OUR VILLAGE

They say it takes a village to raise a child. I always thought of our friends and family as that village; never did I imagine that our village would expand to include perfect strangers, people that we only interact with for seemingly impersonal things like deliveries, canoe rentals and prescriptions.



Take our UPS delivery person, Ed Walters. When Brooke was first diagnosed, and the medication and medical supplies shipments began pouring in, I saw Ed daily. I finally gave him a CRF information pamphlet and explained that these packages were not a result of an online shopping habit (which is what it probably seemed like initially!) but supplies and medication for Brooke. Without hesitation, Ed gave me his cell phone number and encouraged me to reach out if I ever needed anything. I was touched by his gesture, but never thought I would need or want to burden him with our challenges. Fast forward to a weekend this past January, when we were scheduled to leave for vacation but because of our new insurance plan, were hours away from being without Brooke's Procsybi® for several days. While we were finally able to get the medication shipment expedited for Saturday delivery, it wasn't scheduled to arrive before we had to leave. Throw in the fact that a snowstorm was predicted and a signature on delivery required, and we were in a panic. I texted Ed late Friday evening, and he wasn't scheduled to work on Saturday. Even so, Ed responded to me immediately and got in contact with the Saturday UPS delivery person, his manager and his colleague at the distribution center. Within hours I had the contact information for all of them, and they all quickly worked together to locate the package at the distribution center, get it onto the truck and have it delivered early to our home, before the storm and before we were scheduled to leave for vacation. Ed is now my personal hero, and his colleagues are amazing too!



When cystinosis is your life, you spend a lot of time at the pharmacy. Luckily, our local pharmacy is amazing and has become a part of our village! The pharmacists, technicians and staff are so friendly, know Brooke and cheer her on always. Jigar Patel, the owner of the pharmacy, and Walter East, Brooke's personal best friend/pharmacist, are up to date on all of the cystinosis research and always spend time with us when we pick up medications, discussing the current research and ongoing trials. They are available to talk when we have questions, help us navigate the ins and outs of her prescriptions and insurance, and love having Brooke visit the pharmacy. Bellevue Drug Company donates to our fundraiser each year, and Brooke is always showered with affection, attention, stickers and food (there is always a party going on at Bellevue!). We are so blessed to have such an amazing pharmacy in our hometown who know and love Brooke!



By Jill Emerson, Brooke's mom

HAMMONTON, NEW JERSEY



Our village also includes Captain Heber Coltrain, owner of Roanoke Outdoor Adventures. When Clay and the other fishermen decided to hold the 2019 Fishing for Brooke's Cure fundraiser in North Carolina, there were logistics that needed to be addressed for a fundraiser so far from home: how would they transport their personal canoes so far south; if they did rent canoes how could they get them to their fishing location and at dawn, when the fundraiser begins? Clay found Roanoke Outdoor Adventures online, and Capt. Heber jumped in to immediately help. He transported rental canoes to the fishermen at dawn, to a location a significant distance from his business. He then took photos, cheered the fishermen on, and gave them safety pointers on the area. Capt. Heber had never met any of the fishermen before that day and has still not met Brooke. However, based on Brooke's story, he insisted on donating his time and services to our fundraiser and wouldn't even take money to cover his gas for the day. He even checks in with us to see how Brooke is doing and has helped us again for the 2020 fundraiser! We have a friend for life in Capt. Heber!

Dealing with cystinosis isn't easy, as a parent or a patient, but it has certainly helped us realize that even strangers care and want to help. These strangers, who've since become friends, care enough to donate their time, money and energy to Brooke and our family as we navigate this crazy life and work hard to keep Brooke on a healthy track. It really does take a village, and we are so thankful for ours!

READ ABOUT THE
FIFTH ANNUAL
FISHING FOR
BROOKE'S CURE
ON PAGE 67





By Rob Timmons,
Board President,
24 Hours for Hank
SANDPOINT,
IDAHO

FIGHT FOR WHAT YOU BELIEVE IN

I have been fortunate to have known the Sturgis family since the 4th grade and have been including myself in their family events ever since.

Dave and Mary have been as influential in my life as anyone and have always been there to support me and give me guidance when I have needed it most.

Brian and I have been through everything together and most assume we actually “share a brain.” Brian has always been there for me and has never questioned if I needed anything, just how can he help. To say that I owe the Sturgis family is an understatement and I will always be thankful that they have included me as part of their family.

Henry was born in July of 2006, and while everything seemed normal early on, it wasn't long before signs of cystinosis started rearing its ugly face. The confusion and unknown answers were scary and frustrating. Not having a way to help my best friend was the worst feeling and no one seemed to be able to tell us what to do and how to move forward.

In May of 2008, I attended a work event to listen to a speaker named Larry Winget. Larry's message isn't like your typical speakers where they tell you to believe in yourself and “you are good enough.”

In fact, Larry's message may be to some, very offensive and hard to listen to as his whole mantra is that we all need to take ownership of ourselves and stop making excuses for why we can't accomplish our goals. One of the exercises that will stick with me forever is that he had everyone in the audience take out a sheet of paper and write one thing that they

would like to accomplish but that they haven't done yet for one reason or another.

My life changed at that point and I owe it mostly to Larry. After everyone in the room wrote on their sheet of paper what they haven't been able to do, he told us to tear up our pieces of paper and throw them away. His message was clear, it must not be that important to you if you won't take the time required to make it happen. He went on to talk about how people get so consumed in their day to day that they forget or don't make time for what is truly important. That message stung as I opened back up my paper and read what I had written: “To find a way to help the Sturgis family.” How could anything be more important than that after everything they have done for me and my family?

The good news is that I didn't tear up my paper, I took it home and asked my mom and brother to meet me that weekend as I knew I needed help! I needed a plan and I needed to act immediately. It didn't take long after that, after a couple of hours of brainstorming and thinking of a way we could make an impact, 24 Hours for Hank was born. I made some phone calls to a group of friends and family that I knew were also looking for a way to help and we were off and running. The idea for 24 Hours for Hank is simple, cystinosis is a 24-hour disease and treatments are 24 hours a day. We wanted to put people in a place with little or no sleep and push them physically so they could for at least one day, experience a little of what Hank deals with every day.

Things started moving quickly and the support was unbelievable. We decided to host our first 24-hour bike race only 4 months after the initial conversation. We had people reaching out asking how they could help, how they could become part of our events. One thing was very clear and has only gotten stronger every year since. People are looking for a way to help others,

24 HOURS FOR HANK



people are good, and people want to Fight Hank. We have been clear on our foundation's mission statement since day one: "To Find a CURE for Cystinosis." We have hosted three 24-hour bike events, a trail run and are now preparing for our 12th annual 24-hour ski event. We have raised over \$1.5 million in donations and we have been fortunate to be part of something that matters and will someday allow all of us to say we stood up and fought for something we believe in.

I will never be able to thank everyone who has joined our fight and I am beyond grateful for all the new friends, the memories of the past 12 years and for their commitment to beating this disease.

My desire and focus have never been stronger, and I am grateful for what this foundation has taught me. I am full of hope as we head into the new year and I am looking forward to the day when we can announce that we have beaten this horrible disease.





WE ALL BELONG.

By Erin Little, Olivia's mom

PORT ELGIN, ONTARIO, CANADA



Like any parent we all want our children to find genuine and meaningful relationships, so when I received the message asking if Olivia would be interested in being Mary's pen pal it was an easy yes. Of course, it meant explaining what a pen pal was and shopping for all the nostalgic pen pal supplies.

Olivia has friendships with "normal" kids and by normal, I mean kids that don't live with a rare disease. She has playdates and is as normal as everyone else that she hangs out with. We have always instilled in our girls that every "body" is different and that's what makes us all unique. Living in a population of 15,000 people it is highly unlikely that she will find another friend living with cystinosis so we do our best to ensure that she knows that despite her differences, she in fact belongs.

The first letter arrived, and a friendship was immediately started. It was then I realized how important it was for Olivia to find someone that was different like her. Mary went on to talk about all the things that she loved - riding horses, her family, the color purple and she even managed to throw in one thing she hated, getting her g-tube changed. After Olivia read this line, she had a look of shock with a

side of excitement that Mary, like her, has a g-tube. I could physically feel her feeling a bit less alone knowing that Mary was like her. There have been many blessings in their kindred friendship, my favorite being that they talk about everything but cystinosis. In fact, Olivia still doesn't know that Mary has cystinosis like her, we don't want it to be the purpose of the friendship but instead let them build a meaningful relationship around their lives and then one day they will have each other to get through the hard things that we as parents and other friends will never fully understand.

Melissa (Mary's mom) and I have been friends for a few years now, their family Christmas card has hung proudly on our fridge since 2018 and it's beautiful to see our kids organically grow a bond. I can relate to how Olivia will feel one day knowing she has someone "like her" to call a friend. I feel that way about the friendships and bonds I have made over the years attending Day of Hope.

Raising a child with a rare disease has many challenges but we must always remember...

We are all the same. We are all different.
We are all normal. We all belong.

FAMILY, FRIENDS, AND ALL SUPPORTERS OF JPFH AND CRE.



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

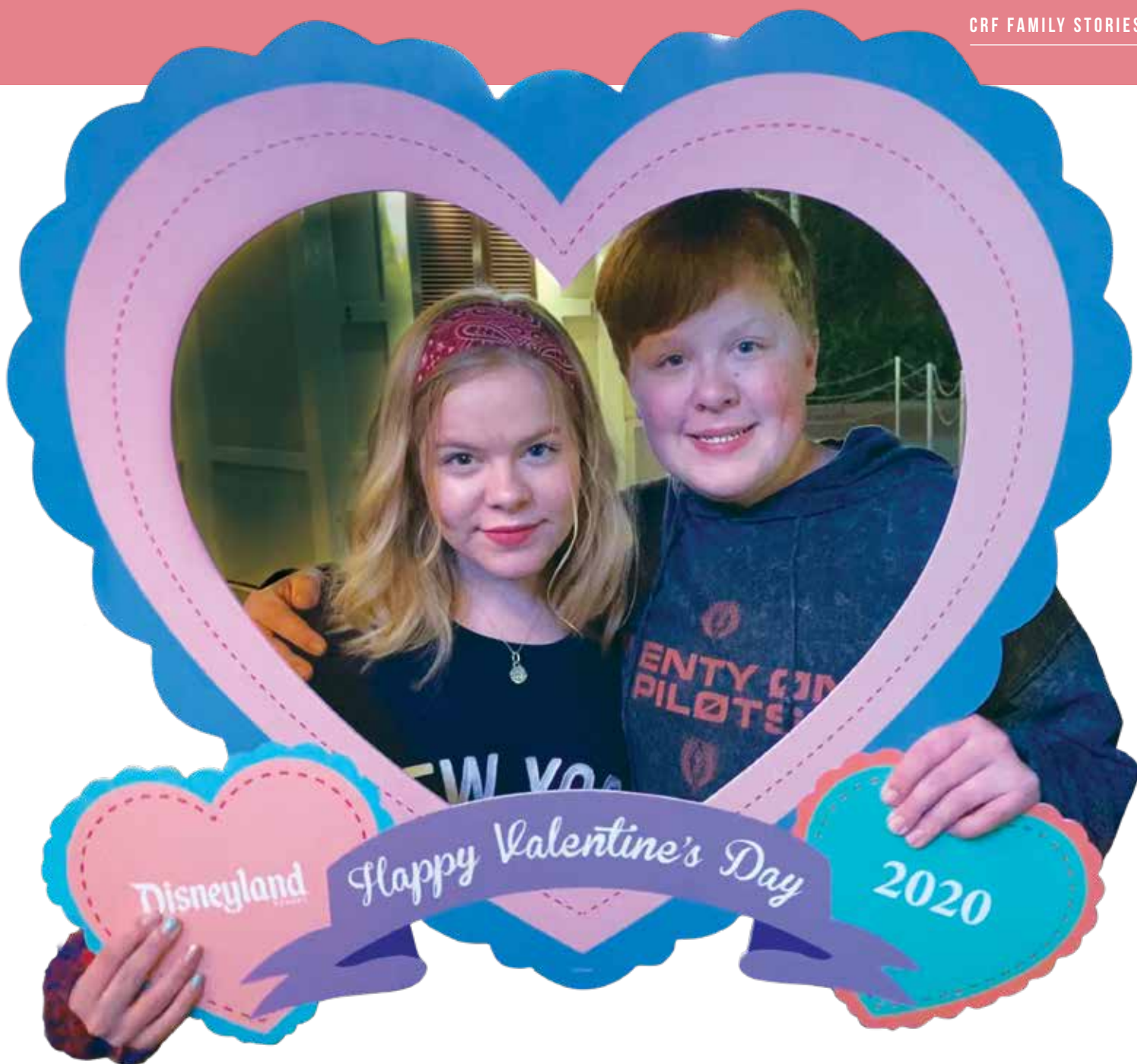
Jenna and Patrick are working hard at their freshman year of high school. The second semester seems a bit more manageable, and they are both quite happy at school. It seems that most weeks, one of them is home for a day, if not both. The twins have about 30% kidney function, and they get very tired very easily. Kevin and I are often conflicted about how much to ask of them each day as they work at their studies and the extra-curricular activities that they are brave enough to add to their days. Patrick joined the chess club at school and continues to enjoy photography. Jenna has served as a tech assistant for the school drama program both semesters this year and enjoys it. She has also bravely chosen to join the swim team at her school. Some days she swims, and some days she serves as team manager and helps out on the pool deck. We are glad to see Jenna as part of a team, enjoying time with friends outside the classroom and challenging herself with something new.

A couple of months ago I took Patrick to the pediatrician for headaches, and it was determined he might have a

condition called pseudotumor cerebri, which means increased pressure in the brain, pressing on the optic nerve and sometimes leading to severe vision problems. The following days saw Patrick missing school and visiting a Neurosurgeon (a six-foot-tall German woman with a thick accent and very little bedside manner), and a Pediatric Neuro-ophthalmologist, Dr. Liu, who is a superstar! Dr. Liu found a lesion on Patrick's optic nerve that seemed to be of concern, so an MRI was ordered. Wouldn't you know, his dental braces interfered with the MRI, and everything in his brain showed up EXCEPT the optic nerve? The next diagnostic step was a lumbar puncture, which confirmed increased cranial pressure and led doctors to remove 12 milliliters of spinal fluid, in an effort to relieve the pressure. We are waiting and watching to understand if his cranial pressure will increase again. If it does, a new medication will be added to his regimen. Another option for pressure relief is the placement of a permanent shunt, which we learned Patrick is not a good candidate for due to very narrow vessels in his brain. Nothing is simple. How does the lesion on the optic nerve play into this? We aren't sure. He will be seen every three months for quite some time. He will need another lumbar puncture. He was so sick after the first procedure as he adjusted to the reduced pressure and missing spinal fluid. It was heartbreaking to see him deal with the pain and discomfort of the procedure, and more heartbreaking to know he will need to do it again. Nothing is simple.

The greatest challenge at this point of our journey is balancing the importance of being in school classes and the critical and time-consuming task of keeping the twins well and tending to doctors' appointments. We encourage the kids to do their best in school while we try to remove as much of the stress





and pressure in their lives as possible. We will defer foreign language for Jenna, and perhaps PE for Patrick, and accommodations and adjustments will be ongoing. Their choice of school classes, semester to semester, may very well affect their eligibility to many universities, but we believe it is most important to create an environment where our kids are able to focus on health and overall happiness. Can you imagine the stories Jenna

**CAN YOU IMAGINE
THE STORIES JENNA
AND PATRICK WILL
BE ABLE TO WRITE
FOR THEIR COLLEGE
ENTRANCE ESSAYS?**

and Patrick will be able to write for their college entrance essays? They'll have that going for them as college looms ever closer!

Finally, we are grateful to Jordan and the team of people who got him to and through the first bone marrow stem cell transplant for cystinosis. What a hero he is to so many people with cystinosis and their families and friends. It is awe-inspiring to consider what is happening in this community. We are forever grateful for your loving support.



RESEARCH. HOPE. CURE.



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A Potential New Treatment for Ocular Cystinosis

An Update Q & A on the SoliDrop Project

Q: Your project is so exciting, please give us a description of the SoliDrop project.

SoliDrop is comprised of two primary components: a reverse thermal gel and drug-loaded microspheres. The gel is a carrier for the microspheres that is administered like a traditional liquid eye drop, before it reaches body temperature and transitions to a stable, pliable, and comfortable solid depot. This depot conforms to the shape of the space beneath your lower eyelid and rests there as drug is released from the microspheres. The microspheres contain dry cysteamine that is trapped within the semi-porous meshwork of the microspheres. The polymer that we make the microspheres from breaks down in water at a controllable and predictable rate, which causes the pores to grow larger and eventually release the entrapped cysteamine.

Q: What is the SoliDrop material made of?

The gel is made of a material called poly-N-isopropylacrylamide containing a small amount of poly ethylene glycol, which is commonly found in laxatives. The microspheres are made of poly (lactic-co-glycolic acid). Poly just means that the material is made of long, repeating chains of whatever name comes after it. So poly (lactic-co-glycolic acid) or PLGA has randomly repeating units of lactic and glycolic acid. The number of those units determines how long it takes the polymer to break down.



BY MORGAN FEDORCHAK, PHD
UNIVERSITY OF PITTSBURGH SCHOOL OF MEDICINE

Q: What is the status of the SoliDrop project?

We have a growing colony of mice (20 pups of varying ages). We would like to get one or two more litters before we start testing the older moms and dads. We are close to having a sustainable colony. We believe that we will know very quickly if the daily SoliDrop is working. This will answer a lot of our remaining questions about dosing and timing. We are hoping to wrap up the mouse studies this year and begin planning the clinical trial alongside that.

The biggest hurdle is resolving human dosing from mouse data and getting a drop to stay put in a tiny mouse eye that has little to no fornix.

Q: How often will SoliDrop need to be administered? Do you think the SoliDrop will be a once a day treatment? Once a week?

I think it will be once per day. The mouse studies will help us begin to understand the dosing and timing somewhat better, as will our larger animal studies in healthy rabbits. The goal is to maximize the therapeutic index—meaning we want it to be as effective as possible while still being really safe and well-tolerated.

Continued on
next page



Q: Can the patient put the drop in at night and sleep with it in?

The idea would be to have one SoliDrop in each eye for a full 24 hours—including during sleep—before replacing with a new one.

Q: Can the patient wear contact lenses and use SoliDrops? Will it cause blurriness?

There is no interference with contact lenses when the gel is properly situated in the fornix (the space beneath the lower eyelid) because it is below the cornea. Used properly, you wouldn't be able to see the gel at all unless you gently pulled on the eyelid specifically to see it.

Q: How will it be administered? Do you need to remove it (yes!) and how?

We are still working out exactly how it will be administered and packaged. But basically you would gently pull out the lower eyelid to form the pocket in the fornix and instill a single drop. There would be some water loss as it transitions over a few seconds and we tend to see best results when the eyelid is used to work the gel deep into the fornix. To remove you would flush with room temperature saline or use a clean finger to remove similar to a contact lens.

Q: How young do you think the patient can be to use the SoliDrop?

I don't know the minimum age (and as with most things involving kids it would definitely depend on a lot of factors!) but my youngest is 5 and I'd feel totally comfortable using this with him.

Q: How quickly do you think the corneal crystals will "dissolve" using the SoliDrop? Do you think all crystals will be dissolved if the SoliDrops are used regularly?

This is the million-dollar question! We are hoping to interpolate this answer from a combination of mouse data (Does a mouse sized dose work? How well and how fast?) and rabbit data (How much drug can we detect in the cornea and how does that compare to what we know about Cystaran™ drops?)

Q: At this point, do you know if there are any side effects from using the SoliDrop?

So far, it's looking like the answer is no. The dose of cysteamine is much lower at any given time than what is in the drops so it should be less irritating. We are going to be evaluating that in our animal studies.



Continued
from Page 43

An Update Q & A on the SoliDrop Project



Q: The current eye drop treatment has a preservative in it which irritates the eye and makes it impossible for some patients to comply with the hourly recommended administration. Is there a preservative in the SoliDrops?

No. We see a seven-fold increase in shelf stability (even at room temperature) with no preservative needed.

Q: Do you expect there will be eye redness as a result of this treatment? Can a patient use Lumify or Visine while on the drug?

I'm not really sure if it will cause redness. The lower dose should help minimize any redness from cysteamine. We have not seen any redness in animal studies (up to 28 days at a time for other drugs) so there won't be any from the gel materials.

Q: In your opinion, how is your proposed treatment superior to the current treatments for ocular cystinosis?

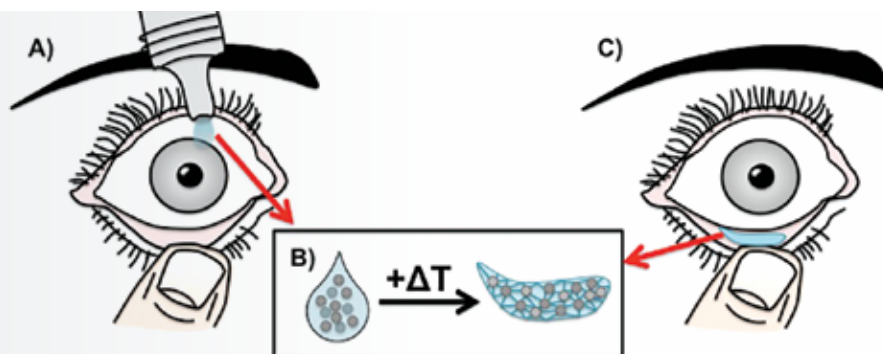
Right now, what we know for sure is that the drops are really stable and the storage is very straightforward compared to Cystaran™ and Cystadrops®. We are hoping to get really good results that show that a lower dose and less frequent administration leads to less irritation and better crystal reduction or elimination (particularly because you would be getting drug continuously in low amounts rather than huge concentrations all at one at discrete times throughout the day).

Q: When do you think we might have a human clinical trial?

If the results look good, I think we could target 2021 to submit information to the FDA. Hopefully, we will have our initial discussions with the FDA this year.

Q: What has CRF support meant to you and your colleagues?

CRF has provided continuous financial support for this project since 2016 and has been instrumental in connecting us to other researchers and patients who have positively influenced the work we are doing in so many ways. They have also allowed us the flexibility to adjust scientifically and administratively when we needed to so that the work can move forward at a good pace. And they are committed to helping us see the project through. It has been really rewarding and helpful to have CRF as more of a partner than just a funding agency. My team feels like we are part of a community and we have a real sense of investment in the project. I think it's a really great model for how rare disease research can be tremendously impactful.



THE IMPACT

OF CRF RESEARCH



AREAS OF
RESEARCH FOCUS
& GRANTS AWARDED
SINCE 2002



Cystine Measurement and Cysteamine Toxicity Study

9 GRANTS

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

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

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



Eye-Corneal Cystinosis Research

9 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



New Drug Discovery Cysteamine, New Medications and Devices

26 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
DE DUVE INSTITUTE,
UNIVERSITÉ CATHOLIQUE
DE LOUVAIN,
BRUSSELS, BELGIUM

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

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



Kidney Research

22 GRANTS

Robert Chevalier, MD
UNIVERSITY OF VIRGINIA,
CHARLOTTESVILLE, VIRGINIA

Pierre Courtoy, MD, PhD
Christopher Pierreux, PhD
DE DUVE INSTITUTE, UNIVERSITÉ
CATHOLIQUE DE LOUVAIN,
BRUSSELS, BELGIUM

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

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

NEW

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

NEW



Skin, Muscle and Bone

13 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,
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
Lee Rubin, PhD
MASSACHUSETTS GENERAL HOSPITAL,
BOSTON, MASSACHUSETTS

NEW



Thyroid

1 GRANT

Pierre Courtoy, MD, PhD
DE DUVE INSTITUTE, UNIVERSITÉ
CATHOLIQUE DE LOUVAIN,
BRUSSELS, BELGIUM

CONTINUED
ON NEXT
PAGE

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

Elena Levtschenko, MD, PhD

UNIVERSITY HOSPITAL,
LEUVEN, BELGIUM

Minnie Sarwal, MD, PhD

UNIVERSITY OF CALIFORNIA,
SAN FRANCISCO,
SAN FRANCISCO, CALIFORNIA

Sihoun Hahn, MD, PhD

SEATTLE CHILDREN'S HOSPITAL,
SEATTLE, WASHINGTON

Eric Moses, PhD

TEXAS BIOMEDICAL RESEARCH
INSTITUTE, SAN ANTONIO, TEXAS



Neurological

15 GRANTS

Angela Ballantyne, PhD

UNIVERSITY OF CALIFORNIA,
SAN DIEGO,
LA JOLLA, CALIFORNIA

Rita Ceponiene, MD, PhD

UNIVERSITY OF CALIFORNIA,
SAN DIEGO,
LA JOLLA, CALIFORNIA

Amy Spilkin, PhD

UNIVERSITY OF CALIFORNIA,
SAN DIEGO,
LA JOLLA, CALIFORNIA

Miriam Britt Sach, MD, PhD

UNIVERSITY OF CALIFORNIA,
SAN DIEGO,
LA JOLLA, CALIFORNIA

Florian Eichler, MD

MASSACHUSETTS GENERAL HOSPITAL,
BOSTON, MASSACHUSETTS

Doris Trauner, MD

UNIVERSITY OF CALIFORNIA,
SAN DIEGO,
LA JOLLA, CALIFORNIA

Aude Servais, MD, PhD

NECKER HOSPITAL, PARIS, FRANCE



Cure Cystinosis International
Registry (CCIR)

1 GRANT

Ranjan Dohil, MD

UNIVERSITY OF CALIFORNIA, SAN DIEGO,
LA JOLLA, CALIFORNIA



Rat Model for Cystinosis

1 GRANT

Francesco Emma, MD

BAMBINO GESÙ CHILDREN'S
HOSPITAL, ROME, ITALY

Olivier Devuyst, MD, PhD

UNIVERSITY OF ZÜRICH,
ZÜRICH, SWITZERLAND

THE IMPACT OF CRYSTINOSIS RESEARCH



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

32 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 GÉNÉTIQUE
MOLÉCULAIRE 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



Cellular and/or Molecular Studies of the Pathogenesis of Cystinosis

51 GRANTS

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

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

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

Liang Feng, 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 Levchenko, 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



Lab Equipment for Cystinosis

3 GRANTS

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

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

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

CRF RESEARCH



GRANTS AWARDS

2019 TOTAL **\$2,617,711**

Francesco Bellomo, PhD,
Principal Investigator
Francesco Emma, MD,
Co-Principal Investigator

Bambino Gesù Children's Hospital, IRCCS,
Rome, Italy

**Ketogenic Diet for
Nephropathic Cystinosis**

\$200,310

TWO-YEAR GRANT

Sergio Catz, PhD, *Research Mentor*
Nadia Zgajnar, PhD, *Research Fellow*

The Scripps Research Institute,
La Jolla, California

**Role of Cystinosin in the Cross-
Talk Between Late Endosomal,
Endoplasmic Reticulum and
Autophagic Functions**

\$150,000

TWO-YEAR GRANT

Stéphanie Cherqui, PhD,
Principal Investigator

University of California, San Diego,
La Jolla, California

**Mechanism of Bone Marrow Stem
Cell-Mediated Therapy in the
Mouse Model of Cystinosis**

\$362,746

TWO-YEAR GRANT



SPRING 2020

PUBLISHED STUDY



**USE OF HUMAN INDUCED PLURIPOTENT STEM CELLS AND KIDNEY ORGANIDS
TO DEVELOP A CYSTEAMINE/MTOR INHIBITION COMBINATION THERAPY FOR
CYSTINOSIS,** *published March 2020 in Journal of the American Society
of Nephrology by Alan J. Davidson, PhD, Teresa Holm, MD, PhD, and
Jennifer Hollywood, PhD, University of Auckland, New Zealand.*



2019 FALL GRANT AWARDS

\$856,488



Pierre Courtoy, MD, PhD,
Co-Principal Investigator (Brussels)
Christophe Pierreux, PhD,
Co-Principal Investigator (Brussels)
Laura Rita Rega, PhD,
Co-Principal Investigator (Rome)

de Duve Institute, Brussels, Belgium
Bambino Gesù Children's Hospital, IRCCS,
Rome, Italy

**Secondary Treatment of Ctns-/-
Mice with Dibasic Amino-Acids:
Safety, Efficacy and Mechanism(s)**

\$ 190,707 TWO-YEAR GRANT

Andrea Del Fattore, PhD, Research Mentor
Giulia Battafarano, PhD, Research Fellow

Bambino Gesù Children's Hospital, IRCCS,
Rome, Italy

**Cathepsin D Inhibition to Rescue
Osteoblast Function in Cystinosis**

\$ 62,975 ONE-YEAR GRANT

Bruno Gasnier, PhD, Research Mentor
Yann Teriz, MSc, Research Fellow

CNRS / Paris Descartes University,
Paris, France

**A Novel Absorptive Epithelium
Defect in Cystinosis**

\$ 75,000 ONE-YEAR GRANT



Robert Mak, MD, PhD, Principal Investigator
University of California, San Diego,
La Jolla, California

**Impact of NLRP3 Inflammasome Signaling
on Fanconi Syndrome and Progressive Kidney
Disease in Infantile Nephropathic Cystinosis**

\$ 348,660 TWO-YEAR GRANT



Reza Seyedsadjadi, MD, Principal Investigator
Lee Rubin, PhD, Co-Principal Investigator

Massachusetts General Hospital,
Boston, Massachusetts

**Clinical, Neurophysiological and Pathological
Characterization of Myopathy and Dysphagia
in Adults with Nephropathic Cystinosis;
Evaluation for Inherent Muscle Resilience
and Regenerative Capacity**

\$ 317,121 TWO-YEAR GRANT

Matias Simons, MD, Research Mentor
Zvonimir Marelja, PhD, Research Fellow

Institut Imagine,
Paris, France

**Elucidation of Tissue-Specific Roles of
Cystinosis in mTORC1 Signaling and
Mitochondrial Metabolism**

\$ 145,000 TWO-YEAR GRANT

Stéphanie Cherqui, PhD

University of California, San Diego, La Jolla, California

Stem Cell and Gene Therapy Clinical Trial

\$ 765,192 FOUR-YEAR GIFT INSTALLMENTS

**CRF
GIFT
AWARD**



SEE 2019 FALL

LAY ABSTRACTS

STARTING ON NEXT PAGE



Secondary treatment of *Ctns*^{-/-} mice with dibasic amino acids: safety, efficacy and mechanism(s)



Pierre Courtoy, MD, PhD, Principal Investigator

Christophe E. Pierreux, PhD, Co-Principal Investigator

Laura R. Rega, PhD, Co-Principal Investigator

DE DUVE INSTITUTE, BRUSSELS, BELGIUM

OBJECTIVE/RATIONALE:

Our CRF-supported research demonstrated that endocytic recapture of ultrafiltered plasma proteins driven by megalin in kidney proximal tubular cells (PTCs; in brief the “megalyn pathway”) is the main cause of cystine accumulation and tissue alterations in cystinotic mice (*Ctns*^{-/-}) kidneys. Ongoing investigations suggest that the megalin pathway can be significantly inhibited, resulting into primary disease prevention, by long-term dietary supplementation with natural dibasic amino-acids already used to treat other genetic diseases, or consumed by body-builders. This new project will assess benefit of secondary protection, i.e. when lesions are already present.

PROJECT DESCRIPTION:

This project is a collaborative effort between Brussels and Roma teams. Cystinotic female mice will be fed from 6 months of age, without or with supplementation by L-lysine or L-arginine in solid diet. Blood and urine analyses every 3 months will monitor inhibition of kidney protein recapture and global function. Mice will be euthanized at 12 months, to assess kidney structure and PTC subcellular/molecular anatomy, with focus on endocytic function. We will also address whether protection only relies on inhibition of endocytic uptake, or further involves anabolic effect via mTOR signaling. To this aim, we will substitute natural (L-amino acids) by their D-stereoisomers, which should equally inhibit megalin but not trigger mTOR signaling. This information will be important for patient monitoring.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

This translational project extends our investigations aiming at harnessing the megalin pathway, as a key source of cystine accumulation in PTCs of cystinotic mice. Our current hypothesis is that high concentrations of dibasic amino-acids in the primary ultrafiltrate can inhibit megalin, thus strongly decrease the recapture of plasma proteins which release cystine upon lysosomal digestion. This upstream approach should be complementary to lysosomal purging by cysteamine. We will also explore whether supplementation impacts on mTOR signaling so as to promote PTC anabolism and prevent autophagy.

ANTICIPATED OUTCOME:

Using a validated mouse model of nephropathic cystinosis, we expect to find that simple dietary supplementation with either L-lysine or L-arginine, natural amino-acid building blocks of human proteins, is safe and can significantly delay progression of kidney lesions and dysfunction, as suggested by preliminary data. If confirmed, this study will pave the way to multicentric pilot testing in cystinotic children.



Impact of NLRP3 inflammasome signaling on Fanconi syndrome and progressive kidney disease in infantile nephropathic cystinosis

Robert Mak, MD, PhD, *Principal Investigator*

UNIVERSITY OF CALIFORNIA, SAN DIEGO, LA JOLLA, CALIFORNIA

OBJECTIVE/RATIONALE:

Infantile nephropathic cystinosis (INC) presents in children as renal Fanconi syndrome in the first year and end-stage renal disease in the second decade despite cysteamine therapy. Fanconi syndrome in children with INC leads to the urinary leak of essential nutrients such as phosphate, calcium, glucose, amino acids and low molecular weight proteins, as well as water. Importantly, Fanconi syndrome is not improved by cysteamine therapy in patients with INC. Our preliminary results suggest that blockade of NLRP3 inflammasome signaling, via both genetic and pharmacological approaches, attenuates Fanconi syndrome and improves renal function in *Ctns*^{-/-} mice, an established mouse model of INC.

PROJECT DESCRIPTION:

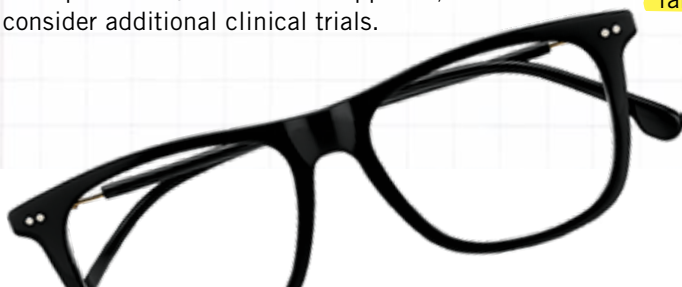
Our hypothesis is that activation of NLRP3 inflammasome signaling negatively impacts renal tubular and glomerular function and leads to Fanconi syndrome and progressive renal failure in patients with INC. We propose to test the pharmacological blockade of NLRP3 signaling as novel therapy for INC. We will investigate whether treatment with the NLRP3 inhibitor (Dapansutrile) and the interleukin 1 receptor antagonist (anakinra) could attenuate the progression of Fanconi syndrome and kidney failure in *Ctns*^{-/-} mice. We will also investigate the long-term safety of these approaches in various vital organs and tissues in *Ctns*^{-/-} mice.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

Dapansutrile is a NLRP3-specific antagonist which blocks NLRP3 signaling. Anakinra is a FDA-approved IL-1 receptor antagonist, which has already been widely used in children with systemic inflammatory diseases. Our hypothesis, if proven, may pave the path for novel therapies for INC. We will aim for a Phase III trial with the results of this preclinical study repurposing FDA approved IL-1 targeted treatment for treatment of INC. When NLRP3 antagonists (such as Dapansutrile) achieve FDA approval, we will also consider additional clinical trials.

ANTICIPATED OUTCOME:

In this proposal, we aim to test the long-term efficacies of these pharmacological approaches on the kidney phenotype in *Ctns*^{-/-} mice. We would also like to assess the safety (potential toxicity) of these long-term therapies on kidney, liver, thyroid and muscle health as a preparation for clinical trials. We expect to observe a significant improvement in renal tubular and glomerular function in *Ctns*^{-/-} mice treated with Dapansutrile or anakinra relative to control mice. Results of this proposal may pave the way for novel therapy for progression of Fanconi syndrome and renal failure in children with INC.





Clinical, neurophysiological and pathological characterization of myopathy and dysphagia in adults with nephropathic cystinosis; evaluation for inherent muscle resilience and regenerative capacity

Reza Seyedsadjadi, MD, *Principal Investigator*

Lee Rubin, PhD, *Co-Principal Investigator*

MASSACHUSETTS GENERAL HOSPITAL, BOSTON, MASSACHUSETTS

OBJECTIVE/RATIONALE:

Major gap in the clinical trials and treatment of weakness (myopathy) and difficulty in swallowing (dysphagia) is the lack of in-depth understanding of the mechanisms of how disease affects muscles and swallowing. We plan to better understand how disease affects muscle and causes weakness and difficulty swallowing using advanced imaging, neurophysiology and pathology techniques.

PROJECT DESCRIPTION:

In this proposal, we aim to better understand evolution of muscle weakness and difficulty swallowing, bringing together the resources of the MGH Neuromuscular Center and the Harvard Stem Cell Institute. We will apply sensitive swallowing biomarkers, such as The Modified Barium Swallow Impairment Profile, and determine the most affected muscle. We will use advanced neurophysiological testing and muscle biopsy in a selected group of patients to better understand mechanisms of muscle weakness and identify potential treatment targets.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

In summary, these experiments will provide better understanding of how disease affects muscles, will help identify better treatment targets and outcome measures and will help evaluate feasibility of stem cell therapy at muscle level to ameliorate muscle weakness and difficulty swallowing in these patients. More sensitive clinical, neurophysiological and pathological techniques can define more subtle clinical changes, characterize adaptive capacity and serve as prognostic outcomes.

ANTICIPATED OUTCOME:

In addition to better characterization of how disease affects muscles and swallowing, we hope to find new treatment targets and examine muscle tissue for potential cell therapy applications. With the recent advancements in the field of stem cell and gene therapy, it is now even more important to better evaluate neurophysiological and pathological characteristics as potential regenerative targets.



CALL FOR RESEARCH PROPOSALS



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 17 short years. Since its inception, CRF has funded 190 multi-year research studies in 12 countries. Our researchers have published 82 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 March, CRF announced \$2.5 million was available for the Spring 2020 call for research proposals and fellowship grants. The grant awards will be announced at the end of June 2020.

In 2019, CRF issued 10 new grants for over \$2.6 million which brings 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 the CRF and advises the foundation on the scientific merits of each proposal.

In 2010, CRF established the Cure Cystinosis International Registry (CCIR) to serve as a hub of information about cystinosis and its complications. Currently, the CCIR is being updated and enhanced to include new questions about recently approved treatments and additional information about the effects of cystinosis. The site will include a Professional Research Portal, so that researchers and scientists who register can access and view de-identified, aggregate cystinosis patient information. The registry will provide essential information that will help us accelerate research and better understand the challenges of cystinosis.



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

LEADERSHIP.
GUIDANCE.
COMMITMENT.

The Scientific Review Board (SRB) is composed of leading cystinosis scientists and experts from around the world. Members are actively involved in the grant-review process, evaluating and analyzing all research proposals that are submitted for potential funding, and advising the CRF on the scientific merit of each proposal.



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THANK YOU FOR YOUR DEDICATION TO
THE GLOBAL CYSTINOSIS COMMUNITY.

SEVENTH INTERNATIONAL

CYSTINOSIS RESEARCH SYMPOSIUM

Sponsored by the Cystinosis Research Foundation

SEVENTH CRF INTERNATIONAL CYSTINOSIS RESEARCH SYMPOSIUM

BY STEPHEN JENKINS, MD, CRF BOARD MEMBER

The seventh CRF International Cystinosis Research Symposium in Irvine, California was a huge success. Scientists from all over the world came together to share their CRF-funded research on cystinosis. Many researchers attended for their first time, and they were warmly welcomed into our community.

As a parent of two children with cystinosis, I was most excited to hear about new potential therapies. This was my third time attending the symposium, so it was exciting to see the progress that scientists have made. Earlier projects that focused on mechanisms and cellular biology have yielded potential therapeutic targets, and now the scientists are testing these therapies in animals.



DR. COWAN

The keynote speaker of the conference was Dr. Morton J. Cowan, from UC San Francisco, who is a distinguished researcher and physician who has pioneered gene therapy for different types of severe combined immunodeficiency (SCID).

CONTINUED
ON NEXT
PAGE

SEVENTH CRF INTERNATIONAL CYSTINOSIS RESEARCH SYMPOSIUM

By Stephen Jenkins, MD
CRF Board Member

Dr. Cowan's presentation was followed by Dr. Stéphanie Cherqui, who gave an update on the first gene-corrected autologous stem cell transplant in a human with cystinosis. So far, the trial participant, who we now all know is Jordan Janz, has done well. He has a good level of corrected cells (vector copy number of 2 as measured in the blood cells) and his white blood cell cystine levels have dropped from 7.8 nmol at the time of transplant to 1.5 nmol three months after transplant. This is an important proof of concept that the blood cells have been repaired, but what about the tissue? Preliminary results based on confocal microscopy of the skin show reduced cystine crystals. His urine output has decreased as well, and he has been able to reduce his phosphorus supplementation, suggesting a possible improvement in Fanconi syndrome. He will return to California as soon as the clinical trial is reopened after the coronavirus threat. At that time they will take biopsies to look at the cystine in the tissue. It was a very exciting talk!



DR. FEDORCHAK

Dr. Morgan Fedorchak gave us an update about controlled-release cysteamine eye drops, which she has been testing in rabbits (without cystinosis) and will now be testing in cystinosis knockout mice.

Dr. Laura Rega shared research she has done with Dr. Francesco Emma on compounds that stimulate TFEB and improve regulation of lysosome turnover and autophagy. When given to cystinosis knockout mice, there was a significant reduction in kidney cystine, even without cysteamine therapy. There also appeared to be improvement in Fanconi syndrome. Next, they plan to test these compounds in cystinosis knockout rats, and hopefully humans in the future.



DR. EMMA

Dr. Pierre Courtoy has previously found that knocking out a certain protein called "megalin" in the kidney prevents cystine accumulation and protects the proximal tubules. He hypothesized that inhibiting megalin with large doses of amino acids would have a similar effect. Early results were promising, however, the long-term studies are less conclusive.

Dr. Paul Goodyer gave an update on a new medication that may treat patients with CTNS nonsense mutations. They've tested it on three patients for three weeks, and there was no evidence of toxicity. The study identified a dosing threshold where WBC cystine levels begin to fall. Additional studies from the lab show that the drug can repair the defect in cellular autophagy as well. From lessons learned in the first phase, the protocol will be adjusted and then expanded at additional sites in the USA.



DR. MAK

Dr. Robert Mak is collaborating with Dr. Hal Hoffman, a prior symposium keynote speaker, on studying the effects of inflammation in cystinosis. They believe that by blocking the inflammasome pathway they can improve Fanconi syndrome.

They have been testing two compounds in this pathway in knockout mice, and they have had improvement in urine output and markers of Fanconi syndrome. New oral medication that blocks this pathway is currently being tested in a phase III trial, so they plan to obtain this compound for additional studies.

Dr. Francesco Bellomo shared research he has done with Dr. Emma on whether the ketogenic diet may be beneficial in cystinosis. They have tested it in knockout mice and found that it reduced polyuria, glycosuria and tubular proteinuria, a sign that it may help Fanconi syndrome. It also reduced inflammation and fibrosis in the kidneys. It is unclear why a ketogenic diet had this effect, so they are going to continue research on this question. The ketogenic diet may lead to ketoacidosis, and people with cystinosis already have metabolic acidosis, so any dietary treatment should require close monitoring and testing in humans.

Dr. Sergio Catz has been studying the effects of cystinosis on chaperone-mediated autophagy for several years, and he tested a molecule in this pathway



DR. CATZ

that may rescue this important function in cells. He created jellybeans containing the compound and fed them to knockout mice, and they had decreased polyuria, phosphaturia and proteinuria, which may mean improvement in Fanconi syndrome. There was no change in cystine levels, so it would likely need to be taken along with cysteamine.

In addition to these exciting updates on possible therapies for cystinosis, we heard talks about bone, muscle and neurologic disease, which are very important issues as people with cystinosis live longer. We heard many talks about the molecular mechanisms of cystinosis, which may yield therapeutic targets in the future. Multiple scientists also shared new models for studying the disease, including genetically engineered rats, zebrafish and stem-cell derived organoids. Dr. Benjamin Freedman was invited to give the keynote address on his work creating polycystic kidney organoids two years ago, and now he is currently working on creating cystinosis organoids which could be used to study the disease and test new drugs.

It was exciting to see the scientists ask each other tough questions and help each other troubleshoot their experiments in a collaborative way.

Every symposium breeds new connections, and I'm excited to see what comes out of these partnerships. I am grateful for all their hard work and I'm amazed at how much CRF and these scientists have been able to accomplish. It gives me and my family so much hope for the future.

SEVENTH INTERNATIONAL

CYSTINOSIS RESEARCH SYMPOSIUM

Sponsored by the Cystinosis Research Foundation

UNITED STATES

KATIE AHNEN - \$900



HADLEY ALEXANDER - \$137,830

ODIN & ATLAS ARDAIZ - \$220



LILY BEAUREGARD - \$5,312

JACKSON BLUM-LANG - \$1,000

OLIVER BRITTEN - \$3,328



CHASE CHODAKOWSKY - \$185



JOSHUA CLARKE - \$10,000

CHARLOTTE COE - \$2,140

MIA COPELAND - \$2,490



BAILEY DEDIO - \$550



HOPE FOR BROOKE -
BROOKE EMERSON - \$26,811



TINA FLERCHINGER - \$23,755



CALEB GOWAN - \$1,417



HOLT GRIER - \$1,295



NICOLE HALL - \$6,522

SHEA HAMMOND - \$3,129



LONDON HARTZ - \$29,682



SAM & LARS JENKINS - \$3,558



JOSIE KANUPKE - \$23,651

SHANNON KEIZER - \$3,260



AARAV KHALASI - \$93,257



HAYDEN KIRCHHOF - \$79,543



JAKE KRAHE - \$22,625



KENZIE LAWATSCH - \$9,188

KALEB LAWSHE - \$1,100



LOLA LONG - \$63,193



PRESTON LUKE - \$800

KEEGAN MANZ - \$4,635

STELLA GRACE MILLER - \$1,530



AIDAN O'LEARY - \$79,360



JENNA AND PATRICK PARTINGTON -
\$185,493



MORGAN PEACHMAN - \$15,660

CHARLIE SIMPSON - \$149,640



HENRY STURGIS - \$200,280



EMMA GRACE SUETTA - \$8,972

PEYTAN TAYLOR - \$150

KADEN THOMAS - \$475

SWEDEN

KAROLIS SCHRODER - \$543

AUSTRALIA

ETHAN FENN - \$1,383



CANADA

SOPHIE'S CHAMPIONS -
SOPHIE BÉTOURNAY - \$34,590

NORA AND ALAN CAMPBELL - \$740



SETH'S CIRCLE OF HOPE -
SETH DEBRUYN - \$22,000



ANDREW CUNNINGHAM - \$22,000

HOPE FOR JAMES -
JAMES FEHR - \$21,875

JORDAN JANZ - \$1,522

AMANDA KUEPFER - \$2,512

MARVELED BY MADDIE -
MADDIE LAWRENCE - \$15,540

MARYLYNN'S ROAD TO A CURE -
MARYLYNN LEPACK - \$5,825



OLIVIA LITTLE - \$83,582



ABBI MONAGHAN - \$14,419

KATHLEEN ROBERTS - \$2,370



GABRIELLE STRAUSS - \$7,103

ALIYAH AND MADELYN WALKER -
\$15,330

NORWAY

DENIS LILLAND - \$600



FAMILY FUNDRAISING *From* AROUND THE WORLD

In 2019, our global community helped raise more than **\$5.1 million** for cystinosis research

Your generosity continues to give hope to those with cystinosis and their families. That hope unites us
We are one step closer to a cure!

IN MEMORY OF

DAVID BRINK - \$1,345

SAMANTHA GROVER - \$3,570

SHANNON PAJU - \$1,500

WESTON TSCHANNEN - \$8,000

FROM FACEBOOK

FAMILIES' FACEBOOK EVENT DONATIONS OF \$83,082 ARE INCLUDED IN THE ABOVE TOTALS.

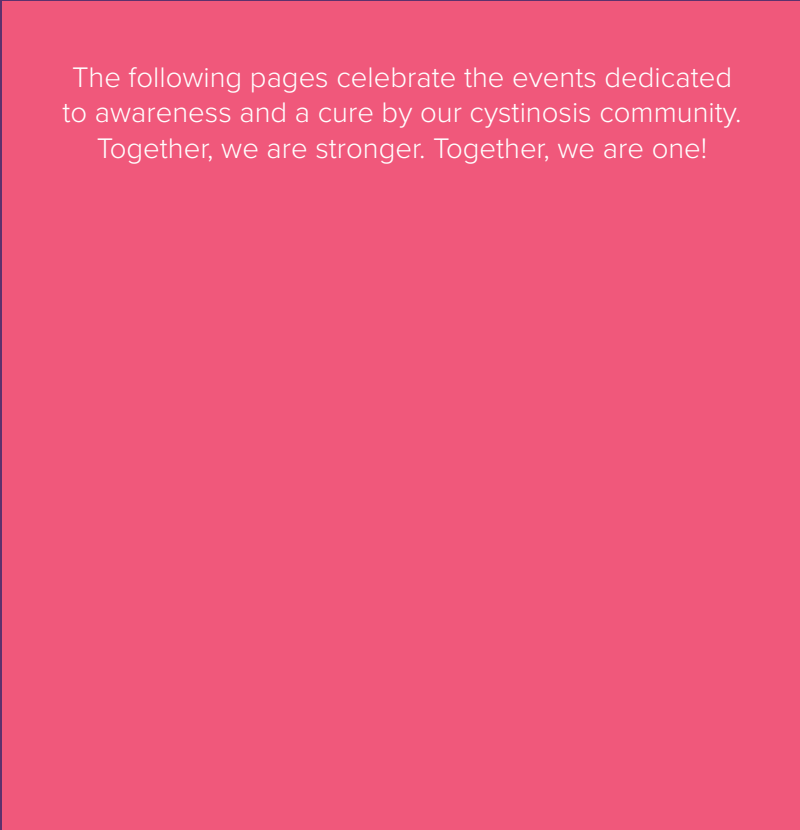


Together We Shine Bright!



TOGETHER,
WE ARE
one

1 PURPOSE. 1 JOURNEY. 1 CURE.



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

1 PURPOSE. 1 JOURNEY. 1 CURE.



Aarav Khalasi – Sacramento, California

AARAV'S TIME TO SHINE

On Saturday, December 28 over 160 people gathered at The Sutter Club in Sacramento, California to attend the inaugural Aarav's Time to Shine Grand Gala fundraiser in honor of Aarav Khalasi.

As guests arrived, they were greeted by Aarav and his parents, Mukund and Minaxi. During the cocktail hour guests bid on a wide variety of special silent auction items. The dinner and event program followed with special guest speaker Teresa Partington sharing the story of her twins with cystinosis, Jenna and Patrick, and the challenges they have faced over the years.



Following the program, a Fund the Mission took place where generous donors donated over \$36,000 in honor of Aarav. Before the event night, hundreds of donations and sponsorships arrived via the CRF website, and as of



now, **the Khalasi family and their amazing community have raised over \$96,300 for CRF and cystinosis research.**

Plans are already underway for this year's event to be held on Saturday, October 3, 2020. CRF is grateful to the Khalasi Family, Aarav's Time to Shine and the generous donors for their commitment to fund cystinosis research for better treatments and a cure!

TOGETHER, WE ARE **One**

Charlie Simpson — New Orleans, Louisiana



GODFATHER RUNS MARATHON IN BROOKLYN, NEW YORK IN HONOR OF CHARLIE SIMPSON

Michael Baumgaertner participated in the NYC Runs half-marathon which was held in Brooklyn in late October. He proudly ran in honor of his godson Charlie Simpson, and with the help of his family and friends, raised \$4,100 on his GoFundMe page. With \$2,500 in matching funds, **Michael raised a total of \$6,600 for CRF and cystinosis research** surpassing his fundraising goal. On behalf of the cystinosis community, thank you Michael!

Braden and Dax Tyner – Milton, Florida

EMILY LEY AND SIMPLIFIED DONATE IN HONOR OF BRADEN AND DAX TYNER

More than 11 years ago, Emily Ley started a stationery and paper business, today known as Simplified. When she learned her friend, Christi Tyner, had two children with cystinosis, she wanted to help and decided to support CRF and cystinosis research. Since that time, **CRF has received more than \$28,380 from Emily's friends and customers in honor of Braden and Dax.** Thank you, Emily, for your commitment to supporting CRF and cystinosis research.



1 PURPOSE. 1 JOURNEY. 1 CURE.

Andrew Cunningham – Langdon, Alberta, Canada

FORE FATHERS ANNUAL JCFG MEMORIAL GOLF CLASSIC

On September 14, 2019 the Eighth Annual JCFG Memorial Golf Classic was held in Langdon, Alberta, Canada. We had tremendous support from our donors and sponsors and a wonderful turnout from our golfers despite our continued recession. Our annual tournament began because four families were impacted by the loss of a father to heart disease and wanted to make a difference. Each of our dads shared a passion for golf, so the obvious choice was to host a golf tournament in their honor. John McCullagh, Conway Cameron, Frank Halluk and Gordon Cunningham aka “The Fore Fathers” became the inspiration of our fundraising efforts.

Andrew is the grandson of two of these great men. **In honor of our ‘Fore Fathers’ we are proud to provide a donation of \$9,000 in support of the CRF.**

Thank you to everyone involved in CRF for all the work you do on behalf of our families. We have full faith in the work being done with Dr. Cherqui and wholeheartedly believe that Natalie’s wish will soon be granted, and all our loved ones’ diseases will go away forever. With much love and appreciation,

Karen McCullagh, on behalf of Fore Father’s Organizing Committee, the McCullagh, Cameron, Halluk and Cunningham Families.



Internet



CYSTINOSIS FAMILIES AND FRIENDS SUPPORT RESEARCH THROUGH FACEBOOK FUNDRAISING



We are forever grateful to all those 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’s never been easier! **In 2019 our Facebook Friends raised over \$83,082 to support CRF.** Additionally, because Facebook doesn’t charge fees on fundraisers for nonprofits, all of that money will go directly to cystinosis research and be put to work as soon as the Spring 2020 grant awards are announced!

To get started, go to this web address: www.facebook.com/fund/CystinosisResearchFoundation

TOGETHER, WE ARE One



Josie Kanupke — Oak Forest, Illinois

JOLLY 4 JOSIE

On Wednesday, December 4 in Tinley Park, Illinois, the Kanupke family hosted their second Jolly 4 Josie event at the Hailstorm Brewery. This year's event started as a low-key gathering but by the end of the night about 100 friends from their community joined them for a Christmas themed celebration in honor of their daughter Josie. Following the raffles, food, desserts and split the pots, guests raised over \$6,800. In conjunction with the event, their friends in the band Rancid hosted an auction for a signed guitar. Flea, from the Red Hot Chili Peppers, was the winner, and he donated the \$9,000 to CRF. **The total for this year's event was \$19,027.** Thank you, Katie and Tom and your community of generous friends for supporting CRF and cystinosis research. Together we are turning wishing into winning in our quest for a cure!



GETTING **Jolly** for Josie

Isaac Andrews — Inver Grove Heights, MN

PINS FOR A PURPOSE

Isaac Andrew's friends and family joined together to plan and support the first annual Pins for a Purpose to raise money for cystinosis research in honor of Isaac. The event was being hosted at Drkula's Bowl in Inver Grove Heights, Minnesota and was to include a moonlight bowling tournament, silent auction and a spaghetti dinner. In early March, there were **125 participants registered and \$1,595 had been raised.** The event was planned for March 21 but due to the COVID-19 pandemic, it was suspended until sometime this summer when it is safe for everyone to come together again.

Isaac's entire network has been instrumental in organizing the event by collecting donations and registering participants to attend. We are so grateful for the support and generosity we have received and cannot wait to get back on track and strike out cystinosis!

*Thank you,
The Andrews Family*



1 PURPOSE. 1 JOURNEY. 1 CURE.

Brooke Emerson — Hammonton, New Jersey

FIFTH ANNUAL FISHING FOR BROOKE'S CURE

On Friday, March 20, 2020, we had another successful Fishing for Brooke's Cure fundraiser. It is hard to believe that this was our fifth event and that it has now been over five years since Brooke's diagnosis.

Our fundraiser occurred just as the US was beginning to understand the scale of the COVID-19 pandemic. This brought an onslaught of widespread health-related concerns and due to the circumstances, two of our fishermen couldn't make it. Nevertheless, our friends Hans and Ralph pressed on and together we social distanced and alcohol hand-wiped our way on a six-hour journey to our fishing destination. We are so honored that even during this time of adversity and uncertainty, our friends and family stepped up to the challenge and we received nearly 200 individual pledges and donations.

Captain Heber Coltrain of Roanoke Outdoor Adventures was kind enough to once again donate his time, services and provide the canoes for our day on the river. We fished from sun-up to sun-down and while the conditions were not what we had hoped for, we managed to catch a combined 164 fish. Thanks to all of our supporters, the fishermen, and of course the fish, we were able to raise more than \$20,400 for CRE, and the donations continue to come in!





CYSTINOSIS COMMUNITY

CALENDAR

OF EVENTS

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.

Monday, July 27, 2020

AIDAN'S ARMY GOLFS FORE A CURE TOURNAMENT IN HONOR OF AIDAN O'LEARY

Forest Lake Country Club, Bloomfield Hills, Michigan

For information contact Katie Emerine at (248) 225-8209 or kemerine33@gmail.com



Summer 2020

PAINT THE TOWN PURPLE IN HONOR OF OLIVIA LITTLE

Port Elgin, Ontario Canada

For information contact Erin.Little@livalittlefoundation.com



Saturday, September 12, 2020

8TH ANNUAL SWING, SHOOT AND LIV GOLF CLASSIC IN HONOR OF OLIVIA LITTLE

Saugeen Golf Club, Port Elgin, Ontario, Canada

For information contact Erin.Little@livalittlefoundation.com



Saturday, October 3, 2020

AARAV'S TIME TO SHINE - GRAND GALA CELEBRATION IN HONOR OF AARAV KHALASI

The Sutter Club, Sacramento, California

For information contact Mukund Khalasi at mukund.khalasi@icloud.com



Thursday, October 22, 2020

ANNUAL SWING GOLF TOURNAMENT IN HONOR OF JENNA AND PATRICK PARTINGTON

Swing Golf Event - Catta Verdera Country Club, Sacramento, California

For information contact Kevin Partington at Kevin.Partington@cushwake.com



Friday, October 23, 2020

7TH ANNUAL HEARTS FOR HADLEY IN HONOR OF HADLEY ALEXANDER

JUMP, Boise, Idaho

For information contact Marcu Alexander at hearts4hadley@gmail.com





LEADERSHIP. GUIDANCE. COMMITMENT.

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Zoe R. Solsby
Vice President

MISSION

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 over \$55 million with 100% of all 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.



Are we
there yet?

Almost!

