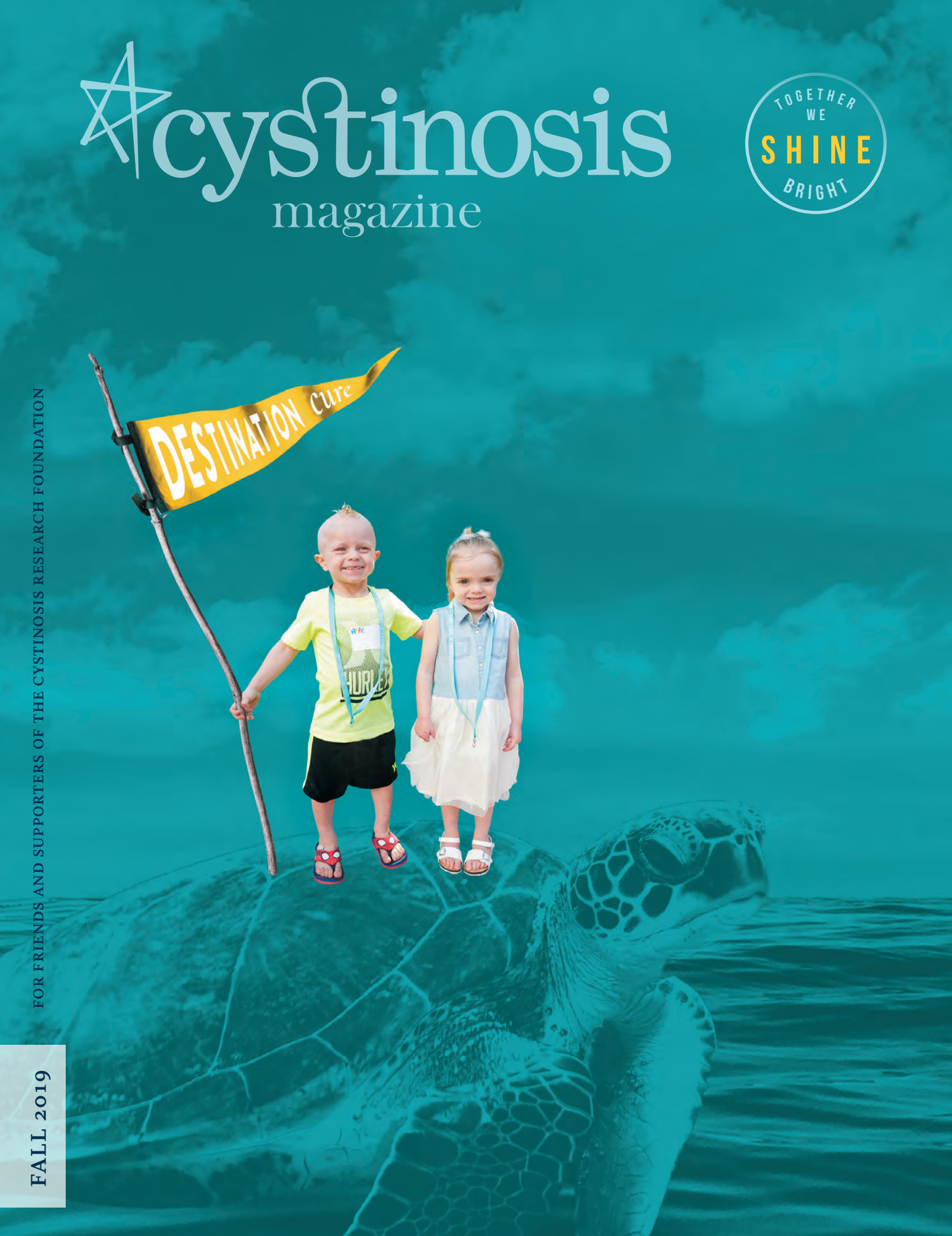
cystinosis magazine

TOGETHER WE SHINE BRIGHT



FOR FRIENDS AND SUPPORTERS OF THE CYSTINOSIS RESEARCH FOUNDATION

FALL 2019

TOGETHER,
WE'RE TURNING

WISHING *into* WINNING!

OUR SIGHTS ARE SET

No potential has limits, and with the recent announcement of the FDA approval for the first stem cell and gene therapy clinical trial for cystinosis, the Cystinosis Research Foundation is grateful and motivated! Our fruitful path of dreams, hope, and positivity are turning into doing, happening and progress!

Together, as we test this new therapy, the CRF community is 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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DESTINATION CURE

FALL 2019

cystinosis RESEARCH FOUNDATION

CYSTINOSISRESEARCH.ORG

NEW ADDRESS
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CHARITY NAVIGATOR ★★★★ Four Star Charity

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 $54 million for cystinosis research in an effort to find a cure.

cystinosis RESEARCH FOUNDATION

WELCOME

CLINICAL TRIALS BEGIN

RESEARCH HIGHLIGHTS

ANNOUNCEMENTS

FAMILY STORIES

FAMILY ADVICE

CYSTINOSIS MAGAZINE IS A PUBLICATION OF THE CYSTINOSIS RESEARCH FOUNDATION

Dear Family *and* Friends

As another year comes to a close, we have so much to be thankful for. We thank and celebrate all of you who have donated to CRF and who have helped us shoulder the day-to-day worries of life with cystinosis.

Although we have had obstacles to overcome and challenges to conquer, we have accomplished great things together. You have been by our side since the inception of the foundation and we are forever grateful. We are a community that is united in our quest for a cure.

We thank our CRF-funded researchers, scientists and clinicians whose tireless work has helped transform the understanding and treatment of cystinosis. CRF has funded hundreds of researchers around the world and as a result, we have created a thriving and synergistic international community of researchers who have dedicated their careers to unlocking the mysteries of cystinosis.

We would not exist without the love and support from families and patients with cystinosis who are the fabric of this foundation. We have woven our unique stories together and we are stronger because of it.

Although Jeff and I started CRF as just one family, we have become a vast group of families and patients from all over the world. It is our strength that has allowed us to thrive and persist.

Natalie's birthday wish, "to have my disease go away forever," was written 16 years ago but it is still our driving force. Her wish became the wish heard around the world and has been a rallying cry for our community. We have created quite a force in the fight against cystinosis; together we have reached milestones and turned "hope" into pioneering clinical trials and new treatments.

CRF RECEIVES PRESTIGIOUS AWARD FOR RESEARCH AND TREATMENT

We are proud to announce that CRF was the recipient of the Global Genes Rare Champion of Hope Award for Research and Treatment. We were honored to accept the award at a special celebration hosted by Global Genes. The Cystinosis Research Foundation has moved mountains and provided hope for those living with cystinosis by funding cutting-edge research that has led to two FDA approvals and new potential treatments. This award belongs to all of us. We are deeply grateful to our cystinosis community, our researchers and all of our donors for their passionate commitment to finding better treatments and a cure for cystinosis.

THE STEM CELL AND GENE THERAPY TRIAL HAS STARTED!

We began funding Stéphanie Cherqui, PhD at UCSD in 2006 and have issued grants totaling over $5.4 million for her research. Since that time, the seed money we provided has been leveraged by other funding agencies, including the California Institute for Regenerative Medicine (CIRM) and National Institutes of Health (NIH). It has not been an easy road to FDA approval, but Stéphanie never gave up, remaining focused on her research—and now the treatment is a reality. We are making history as the first patients are transplanted.

The Phase I and II trial of the genetically-modified autologous stem cell transplant was approved by the FDA for six patients 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 cystinosin gene. These stem cells (without cystinosis) are then transplanted back into the patient. We hope that this one-time treatment will stop the progression of cystinosis and possibly repair some of the damage caused by cystinosis.

We received an overwhelming number of inquiries about the trial from adults with cystinosis. So many adults expressed their thanks for the opportunity to be part of the trial and their gratefulness to CRF and all of you who have funded the research that has given them renewed hope for a brighter future. We were humbled by their responses, which were heartfelt and emotional. Although the recruitment process has been slower than we anticipated because of FDA requirements, **we are moving forward and hope and pray this treatment will be the cure we anticipate it will be.**

The clinical trial is a reality because the first patient, Jordan Janz, was transplanted on October 7, 2019 (see page 16 for Jordan's story). In fact, by the time you read this magazine, we hope that Jordan will be on the road to a full recovery. Jordan is our hero, a true pioneer in our community. There are no words to express our deep thanks and gratitude for the gift of hope that Jordan is giving all of us by volunteering to be the first patient transplanted.

ONGOING RESEARCH PROVIDES HOPE FOR A BRIGHTER FUTURE

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, are painful to administer. CRF is funding two important studies that could revolutionize eye treatment for cystinosis.

SoliDrops - Morgan Fedorchak, PhD

We are more excited than ever by 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 thermosresponsive hydrogel that contains spray dried, cysteamine-loaded microspheres, which turn 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.

CRF has committed to fund Dr. Fedorchak's work throughout the next critical phases which include final formulation, animal studies and the IND (Investigational New Drug) application.

NanoWafer, Inc.

The nanowafer is a unique potential treatment for corneal cystinosis. It is a wafer that can be loaded with medication and placed in the eye where it dissolves but leaves medication in the eye for hours, potentially making it a once-a-day treatment. The nanowafer would be a superior treatment to the current hourly eye drops, which are onerous and painful to take. NanoWafer, Inc. is finalizing the formulation, manufacturing, testing methods, clinical attributes and preparation for production scale up. There has been progress by our team of experts, yet risks and challenges remain. Our plan is to find a partner to work with us to take this technology through the FDA.

A COMMITMENT TO NEW RESEARCH

Given the complexity of cystinosis and the reality that it affects every cell in the body, we must remain vigilant in funding new research. We don't know what new discovery or potential treatment might work for cystinosis, so we maintain a broad portfolio of research projects to ensure we are always in front of the research and are supporting the best researchers at world-renowned institutions. Although the stem cell and gene therapy clinical trial has started, not every patient will choose the treatment; nor will all qualify. We must, therefore, remain committed to funding current and future research.

SEVEN NEW GRANTS TOTALING $1,761,223 ISSUED TO DATE

We are pleased to announce that in the first half of 2019, CRF issued seven new grants totaling $1,761,223 for cystinosis research. The grants were awarded to researchers in the United States, France and Italy. The seven recipients of the Spring 2019 grant awards are listed on page 56 along with lay abstracts of their study.

Since 2003, CRF has funded 187 multi-year research studies in 12 countries. Our researchers have published 80 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. We have listed the current open clinical trials that are seeking patient volunteers on page 10.

We fund new grants twice a year, thereby ensuring that donations to CRF are always funding research. We have built a strong, research-driven foundation that has accomplished major milestones, including two FDA approvals. We can promise that there will be many more life-changing treatments and discoveries in the near future. You have been with us every step of the way, supporting us and encouraging us. We could not do this without you.

TOGETHER,
WE'RE TURNING
POSITIVITY *into* PROGRESS!

THE POWERFUL EFFECT OF CYSTINOSIS RESEARCH ON OTHER DISEASES

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. We are soaring to new heights and we are helping others along the way.

WE ARE FOREVER GRATEFUL

These are exciting times for us! The start of the stem cell trial has brought so much of our work full circle. As the first patients are treated, we will remain cautious and hopeful that the treatment will be the cure. We are grateful to all of you who never wavered in your support of our efforts to find better treatments and a cure for cystinosis. You believed in us and we thank you for that gift of support.

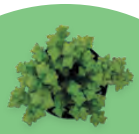
There isn't a day that is healthy for our children and adults with cystinosis. There is not one day that our children can miss taking their medications. They suffer every single day from the side effects of the treatments and the disease. As a mother of an adult with cystinosis, I often feel a deep sense of sadness because my beloved daughter has lived with this horrific disease for 28 years. I find calm and peace knowing that we are working as fast as we can to find a cure for Natalie and all of the other children and adults with cystinosis whom I love like they are my own children.

Thank you for loving our community, for letting us lean on you while we find our way to the cure. We are blessed by your dedication to our community and our mission. You have lifted us up when we have been challenged and you have brought us joy and given us encouragement when we have needed it the most.

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

With heartfelt thanks and gratitude,

Nancy & Jeff



I am eager to see what unfolds over the next couple of years as we move through the clinical trial.

Dear Family and Friends,

I cannot believe 2019 is almost over. It has been an incredible year with lots of hope and faith for cystinosis. I am so looking forward to 2020 because it will be the year the cystinosis community will reach a huge milestone – curing cystinosis. I look forward to the day where no one has to worry about raising money to find a cure and no patient has to worry about his or her future.

I am still in a state of disbelief that the stem cell and gene therapy clinical trial is happening! It is surreal and incredible to know that this is happening so soon and for such a small, rare disease. All of the dedication, drive and hard work my mom, the cystinosis community, and the researchers and doctors have done over the years has finally paid off.

I am so excited to see how the first patients do in the trial; they are courageous and brave to lead the community. I hope that I will be chosen for the trial in 2020 – it would mean my wish has become a reality! I am nervous and anxious, but also incredibly confident that the work Dr. Cherqui and her team have done will result in a positive outcome for those transplanted. I am thankful for Dr. Cherqui's commitment to cystinosis research and for her tireless efforts that have resulted in the recent FDA approval. I believe that the treatment will work and we all will be cured one day soon. I am eager to see what unfolds over the next couple of years as we move through the clinical trial.

On a personal note, I have been working hard at my new job at CASA (Court-Appointed Special Advocates) and I am about to realize my one-year anniversary in October. In my position, I support, recruit and retain volunteers who work with children in the foster care system. The volunteers work one-on-one with the children and advocate for them in juvenile court.

I have learned how to work with many different personalities and have been able to use my social work skills on a daily basis. I love working at CASA – it has forced me to step out of my comfort zone and challenges me personally. I learn something new every day and know that I am making a difference in some small way in our community. I look forward to continuing to grow in my position and to learn how to best advocate for children in the foster care system.

As always, I am beyond thankful to my parents, doctors, researchers and the entire cystinosis community for dedicating their time, funds and efforts to finding a cure for cystinosis. Words will never be able to express what it means to me to have this community by my side fighting for a life free of this disease.

I have hope that I will live longer now, and, because of that, I am extremely grateful to Dr. Cherqui and this community for what they have done for not only me, but all the other children and adults with cystinosis.

Thank you for never giving up and making my wish come true.

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

cystinosis RESEARCH FOUNDATION

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 187 grants in 12 countries.

CRF has raised more than $54 million, with 100% of your donations going to support cystinosis research. CRF's efforts have changed the course of cystinosis and given new energy to its investigators and scientists. CRF's commitment to research has given hope and promise to the global community of cystinosis patients and their families.

CHARITY NAVIGATOR ★★★★ Four Star Charity

WE CELEBRATE OUR CRF COMMUNITY AND ARE GRATEFUL EVERY DAY FOR YOUR SUPPORT.

We're turning wishing into winning with more new clinical trials than ever before. It is because of you that CRF has been able to fund extraordinary researchers across the globe.

THANK YOU!

You have changed the course of cystinosis!

Since 2003

CRF Has Raised More Than

$54 Million

For Cystinosis Research

100%

Of Your Donations Directly Support Cystinosis Research

CRF Researchers Have Published

80 Articles In Prestigious Journals

and Received

187

Multi-Year Grants

In 12 Countries

In 2019

CRF Funded 7 New Research Grants

In 3 Countries

Totaling More Than

$1.7 Million

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.

TOGETHER WE **SHINE BRIGHT**

cystinosis RESEARCH FOUNDATION | TOGETHER WE SHINE BRIGHT

TOGETHER, WE'RE TURNING

RESEARCH · HOPE · CURE

DREAMING into DOING!

CRF STUDIES RESULT IN

CLINICAL TRIALS

HERE ARE JUST A FEW OPEN CLINICAL TRIALS THAT ARE LEADING TO BETTER TREATMENTS AND A CURE FOR CYSTINOSIS

FDA-APPROVED HUMAN CLINICAL TRIALS FOR STEM CELL AND GENE THERAPY TREATMENT FOR CYSTINOSIS

UNIVERSITY OF CALIFORNIA, SAN DIEGO
STÉPHANIE CHERQUI, PhD

1

This study is a huge milestone for CRF and the cystinosis community. Stem cell and gene therapy holds the hope that one day cystinosis will be cured and will be a disease that our children "used to have." The phase I/II human clinical trials will be held at UC San Diego Health in San Diego, CA. The recruitment individuals must be 18 years or older and meet specific eligibility requirements to participate. The subsequent phases of the clinical trials may include children; however, timing and approvals of this have yet to be determined. The trials are available to individuals worldwide, though priority will be given to U.S. patients.

The complete Clinical Trial Inclusion and Exclusion Criteria are available on our website *www.cystinosisresearch.org/fda-approved-clinical-trial-for-cystinosis.*

If you are an adult with cystinosis and are interested in participating in the trial or would like more information, contact Laura Hernandez, *lhernandez@ucsd.edu*, or visit the CRF website – FDA Approved Clinical Trial.

ELOXX PHASE 2 CLINICAL TRIAL OF ELX-02 FOR TREATMENT OF CYSTINOSIS IN MONTREAL, CANADA

2

MCGILL UNIVERSITY
HEALTH CENTRE, CANADA
PAUL GOODYER, MD

It is exciting to know that CRF-funded research is shaping future treatments for cystinosis. Dr. Paul Goodyer, who is conducting the study, was funded by CRF for many years, and his work resulted in this new clinical trial that will take place in Montreal, Canada. Dr. Goodyer and his team at McGill are looking for cystinosis patient volunteers with nonsense mutations who are over 18 years of age with stable kidney function (eGRF>40 mL/min/1.73m2).

Those interested and those who would like more information should contact Murielle Akpa, PhD, by phone (*514-412-4400 ext. 22953*) or by email (*murielle.akpa@muhc.mcgill.ca*). It should be emphasized that subjects will spend six weeks in Montreal, Canada, and will have to discontinue cysteamine during the trial. The recruitment letter is available on our website at: *www.cystinosisresearch.org/clinical-studies-volunteers*.

3 SLEEP AND MEMORY STUDY

UC SAN DIEGO, DORIS TRAUNER, MD

The more we learn about cystinosis, the more we can help our children and adults, and improve their quality of life. Dr. Trauner and her research group at UCSD are conducting a study to evaluate the relationship between sleep, memory and thinking in adults with cystinosis. Dr. Trauner is looking for adults (over 18) with cystinosis to participate in an overnight sleep study and cognitive testing focusing on memory.

Participation involves completing questionnaires, sleeping overnight in a UCSD sleep laboratory and performing tests of memory and thinking. The study will pay for travel and meals, and participants will receive the results of both their memory and thinking tests, as well as their sleep assessments. Volunteers will also receive $150 in compensation for their time. If any problem is found in the sleep study, we will send the results to your primary care physician. For more information, please contact Jennifer Crowhurst at *jcrowhurst@ucsd.edu* or Tammy Vu at *858-822-6700*.

4 RESEARCH STUDY – THERAPEUTIC STRATEGY FOR CYSTINOTIC NEPHROPATHY WITH iPS CELLS

UNIVERSITY OF WASHINGTON
BENJAMIN "BENO" FREEDMAN, PhD

The goal of our CRF project is to use human mini-kidneys as surrogates for patients, to explore the potential of kidney regeneration, gene therapy and drug discovery for cystinosis. We are seeking to recruit patients with cystinosis and turn their urine into stem cells and kidney grafts, which we will test for transplantability. There is no obligation. Study participation is on a purely volunteer basis and is free of charge without compensation. If you are interested in joining the study, or have questions about it, please approach Dr. Freedman at *benof@uw.edu* or Nancy Stack at *nstack@cystinosisresearch.org* for more information.

NEW PHASE IS A SPECIAL MOMENT

by Dennis Arp

As the much-anticipated stem cell trial begins, Dr. Stéphanie Cherqui shares in the excitement while also sticking to the process.

Dr. Stéphanie Cherqui is used to waiting. Over her years of tireless research pursuing a cure for cystinosis, she has endured countless hours anticipating the results of lab analysis, and untold days waiting for news on the future of research studies.

Most recently, she spent a month on pins and needles before getting a call from the U.S. Food and Drug Administration with a verdict on her application to begin a trial of stem cell therapy. When she finally heard her phone ring, she feared the worst.

"People say that if the FDA calls (instead of emailing), it's not a good sign," recalls Dr. Cherqui, associate professor of pediatrics at the University of California, San Diego. "But the project officer said, 'I'm happy to tell you that your trial has been approved.' He kept talking, but I stopped listening – I was just so happy. I told him at the end, 'You'd better email me the details, because it still doesn't seem real.'"

For far too long, cystinosis patients have persevered through a host of grim realities. So it's extra special that this moment turned out to be positively real.

"It was amazing news," Dr. Cherqui says. **"The kind of news you live for."**

Tears flowed and celebrations followed. But then Dr. Cherqui and her team quickly set about the business of making the most of their opportunity. The Phase I/II clinical trial of therapy using stem cells from bone marrow was kicked off in July, allowing the life-changing potential of Dr. Cherqui's gene research to really hit home for cystinosis patients and their families.

Thanks to funding from the Cystinosis Research Foundation as well as the California Institute for Regenerative Medicine, the project was in place and

Stéphanie Cherqui, PhD and her family.

the team was ready to roll. Word went out to the cystinosis community that the trial had been approved and six adult patients were needed. More than 20 responses poured in and were evaluated by a consortium of 15 members, 13 of them cystinosis physicians. The first patient has been enrolled, and the initial transplantation procedure occurred on October 7th. Meanwhile, the enrollment process remains open.

The evaluation process takes time and involves robust testing to ensure that trial participants have the best chance of success, Dr. Cherqui says.

Because the Phase I/II trial is primarily a safety study that marks the transition from animals to humans, only adults are eligible. As an extra precaution, study patients will receive the therapy one at a time, allowing for at least one month of evaluation after transplantation before the next patient can be enrolled in the study and have their own stem cells corrected to introduce a functional CTNS gene.

The treatment has rescued mice from cystinosis and improved their corneal cystinosis as well. What's more, a single stem cell transplantation prevented thyroid dysfunction in mice.

Dr. Cherqui recognizes and shares the excitement of cystinosis patients and their families that the therapy might represent the long-awaited cure for cystinosis. But she cautions that there are still many hurdles to clear on the road to final approval of autologous stem cell and gene therapy.

"This is a first in human therapy for cystinosis," Dr. Cherqui says. "That's why it's hard to say when or if we will see signs of efficacy."

However, Dr. Cherqui can say that the team "is adapting very well – working hard every day."

In all, the project involves as many as 40 team members, including 12 physicians – nephrologists, endocrinologists and every other kind of pertinent organ specialists.

"It's a very impressive team," Dr. Cherqui allows.

For each patient, the process will begin with tests to evaluate the clinical status of all organs. To be eligible, patients must be at least a year out from a kidney transplantation and be free of any infection, which could endanger them during the cell-transplant process.

After trial participants are admitted to the hospital at the University of California, San Diego, they will have their stem cells harvested and shipped the same day to the University of California, Los Angeles, where Dr. Donald Kohn and his team will perform the gene-correction procedure. That process takes about three days and includes testing to ensure the corrected cells are safe for transplantation.

"It's a delicate process to make room in a patient's bone marrow for new stem cells to take the place of old ones," Dr. Cherqui notes. But if all goes well, the new gene-corrected cells will live and multiply in the marrow, becoming a source of healthy stem cells for the rest of a patient's life.

For each patient, the transplantation procedure will require a hospital stay of three to four weeks.

"This is the time when the patient is most at risk because of a compromised immune system," Dr. Cherqui says.

Once patients are discharged, they will need to stay in the San Diego area for an additional two months of weekly testing "to make sure everything is fine," she adds.

In addition to praising the dedication shown by the dozens of members who make up the project team, Dr. Cherqui voices admiration for the trial's first patient, who assumes "a lot of risks," she says.

"If this trial is a success, the first patient and the ones who follow will be a major part of that success."

Together, the trial patients and other contributors form "an amazing partnership," Dr. Cherqui says.

"We all just want to say a big 'thank you' to everyone in the cystinosis community for all the support," she enthuses. "We are in a good place now, with an expectation that we will see efficacy, while at the same time knowing there are still a lot of unknowns. We will see where we stand in a few months. It really is a one-step-at-a-time process."

I walk 100kms every year to raise money for kidney disease. We are encouraged to display our "why."

JORDAN JANZ IS LEADING THE WAY

by Barbara Kulyk, Jordan's mom

Our journey with cystinosis started like most. Jordan was born perfectly healthy in the high percentiles, happy and loved. At three months of age his growing stopped but because of his growth early on this was dismissed time and time again. By five months of age he began projectile vomiting and by seven months was down to the third percentile. I took Jordan to the pediatrician where I used to live, and he was immediately concerned and hospitalized Jordan. In eight days, Jordan was diagnosed with cystinosis, confirmed by the discovery of crystals in his eyes.

A g-tube was placed at eleven months due to the excessive vomiting and this gave me control over his nutritional intake and a sure way to combat dehydration. We lived three and a half hours away from the closest hospital that could treat Jordan if needed.

He started school and struggled with reading and fell behind quickly. I found that the education system at the time wasn't open to finding the proper learning disability diagnosis. A diagnosis was so important because otherwise, we wouldn't know how to teach Jordan the way he needed to be taught. I had him tested independently and he repeated grade two. Although this was hard at the time, he stopped thinking he couldn't learn and soon gained some confidence. As time went on Jordan was given tremendous support at the school; he formed positive relationships with his teachers and eventually obtained his diploma.

He had good friendships with his peers but always understood that he couldn't do the things that they were doing. Sleepovers were not an option, sports left him exhausted, school was still a challenge and by the time homework was done he didn't have a lot of free time or energy. Sleep was always interrupted with the need for six-hour medications, bathroom breaks and the incessant alarm going off from his night feed machine.

At the age of 10, he was enrolled in the 12-hour medication trials in Chicago. I felt that as long as Jordan was little, I could give him his medications on time, but would he continue and be able to continue the strict regimen when he became an adult and lived on his own? I had older kids that were teenagers and it made me wonder: if he got home after midnight after being out with friends would he be able to get up at six a.m. and do his medications on time. I felt that if he was successful on 12-hour cysteamine this could make it easier for him to manage and remain compliant.

Fast-forwarding a few years, the 12-hour cysteamine significantly improved Jordan's quality of life. His vomiting decreased, his appetite improved, we removed his g-tube at 13 and his confidence soared enough for him to take

My family, my daughter Brittany and her husband Brody, their two kids Bridgette and Burkley, Jordan, Nathan and his wife Megan, myself, Jeff and our two little ones Victoria and Easton and my mother Leona.

October 7, 2019, Luke (Jordan's dad), Nancy Stack, Stéphanie Cherqui, PhD, Jordan, Barbara and Jeff (Jordan's mom and stepdad).

lifeguard training and be employed annually at the local pool. I am not saying that the 12-hour medication was a miracle drug, but for Jordan, it started a chain reaction to a better life. He could sleep through the night which gave him more energy and more of an appetite, he started working out with a trainer and joined the local volleyball and baseball teams and swam every chance he got.

Jordan made many meaningful relationships with the people he has met in Chicago and was asked to speak at the CORD (Canadian Organization for Rare Diseases) conference in Toronto in 2017.

He has learned to make the best out of bad situations and to count his blessings. When having a bad day, and we all know there are plenty of those, he takes a break.

Cystinosis is heartbreaking and has given all of us tremendously difficult situations to navigate without a compass. My husband and my five children have become a very close, solid family and we are always there to support each other. I have missed many of my other children's important events to be with Jordan. The mother's guilt is always there. But we do what we can and now Jordan has grown and is guiding this disease his way and now I follow him.

He lives on his own, nine houses away (yes, I counted), and works full time; he cooks the meals he needs and never misses his medications. I know this because I count those too.

He recently decided that he wanted to apply for the stem cell transplant trial to see if he could be a candidate. Turns out he was! With many preparations at work and home Jordan and I have taken a leave of absence and will be in San Diego for a few months. My husband Jeff, my mother and my older children and their spouses are taking care of my little children while we are away.

It has taken the Cystinosis Research Foundation; the team at Cherqui Labs; the generous people that employ us; our community that is cooking for my family at home and face timing my daughters volleyball games to me; the nephrology clinic at Alberta Children's Hospital; and so many people to get us to San Diego for Jordan to be admitted.

Jordan had his transplant and now we wait. I wholeheartedly believe that this will cure him. But of course, you have to entertain the idea that it may not.

I am immensely proud of Jordan for trying and for fighting for a better life. After all, that is what hope is: a desire to be in a better place.

cystinosis RESEARCH FOUNDATION | TOGETHER WE SHINE BRIGHT

RARE CHAMPIONS OF HOPE CELEBRATION

The Cystinosis Research Foundation was recognized by **Global Genes** as a Rare Champion of Hope Awardee for Research and Treatment. Together, we celebrate with all of the champions of hope for rare disease! We are deeply grateful to our community, our researchers and all of our donors for their steadfast commitment to finding better treatments and a cure for cystinosis. Thank you, Global Genes, for recognizing CRF and the cystinosis community for our research accomplishments!

RESEARCH · HOPE · CURE

TOGETHER,
WE'RE TURNING

HOPING into HAPPENING!

Undeniable PROMISE

By Kathleen Roberts

KAMLOOPS, BRITISH COLUMBIA, CANADA

In 2001, when I was 12 months old, I was diagnosed with cystinosis. My diagnosis was pretty typical: hospitalized for three weeks with failure to thrive, including weight loss; vomiting at least six times a day; and a crazed obsession with drinking water. When I was first admitted to the hospital, I shocked the nurses by guzzling two liters of warm, unflavored Pedialyte. Most kids won't even touch this stuff.

As my parents describe it, and from pieces that I recall, those first few years were tough, but full of quirky stories. The time our garbage truck wouldn't take our bin because it was too heavy. It was only wet diapers! When my entire family celebrated because I ate half a strawberry, as for years I very literally didn't take anything by mouth (hello, g-tube and pump full of Pediasure!). When I was a bit older and my parents took me out for dinner to a fancy restaurant and had nice meals themselves, but only gave me free pats of butter while the entire restaurant stared. At that point, butter was the only thing I would eat. I also vividly remember the snowman-decorated vomit bucket that truly went everywhere with me for the first 10 years of my life. My gag reflex was extraordinary; I would vomit anytime or place.

For as long as I can remember, I knew and could pronounce the word cystinosis.

I also remember times when other kids expressed how they thought my medical issues were a big deal. I didn't get where they were coming from. It wasn't a big deal to me. My parents were always very open about cystinosis. For as long as I can remember, I knew and could pronounce the word cystinosis. I knew I would need a "new kidney" someday and that I had to take my meds or I would slowly get sick and die. This may seem morbid for a little kid, but it was just the facts and it didn't bother me. I actually think it positively aided me in accepting when I eventually needed a transplant at 15, and being at peace with my disease later on.

Early on, and for much of my later childhood, we could have never imagined the advanced research that CRF would facilitate and the supportive cystinosis community that would continue to evolve. In 2001, my mom was sent a VCR tape about cystinosis from another family. That was how she learned about life with a cystinosis child. Now there are blogs, Facebook and care packages. In 2001, I was started on Phospho-cysteamine, a liquid that was so foul smelling it needed its own fridge. Cystagon® wasn't available in my province of British Columbia yet. My parents had to fight for it. Now there is Procysbi® and potentially a cure. In 2001, the first thing my parents learned about cystinosis was that I wouldn't live past 10 years old. Now, it's 2019 and I've graduated high school, started my second year of a university nursing degree (UBC) and, if the stem cell trial works out, I have the potential to live a full "normal" lifespan.

Eighteen years may not seem like a lot, and in the grand scheme of things, it isn't. But for advancements in cystinosis it has been. Thanks to CRF, developments in the understanding and treatment of cystinosis and promising outcomes in the search for a cure have occurred during those short 18 years since I've been diagnosed.

I've never believed I needed to be "fixed." Cystinosis has given me equally as much as it has taken. But to live a life without the complications and looming presence of this disease would be marvelous.

Charlie and his big brother, Beckett.

Charlie's Adventure

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

On June 21, 2019 – just one week after his first birthday – our younger son, Charlie, was diagnosed with cystinosis. Since then, we've experienced the surreal and heart-wrenching journey of our baby boy fighting this terrible disease. Our story is a familiar one to many cystinosis families – our poor, tiny child had severe GI issues and wasn't growing. We spent a long, difficult time in the hospital to find out why. Perhaps toughest of all, we learned of the progressive, life-threatening reality of cystinosis weeks after we thought all of the truly scary possibilities had been tested and ruled out.

Our battle with cystinosis has just begun, and we've been told that this time period is one of the most difficult. We often struggle – along with Charlie's wonderful team of doctors and therapists – to find the right balance of medications to treat and slow the progression of the disease (requiring weekly blood draws and frequent appointments, emails, phone calls and texts). We're also constantly working to achieve the best timing and volume for the formula and supplemental feedings that will give Charlie enough nutrition, but won't trigger vomiting several times a day (currently our most frustrating and discouraging symptom). Above all, we're figuring out how this disease will factor into our everyday life.

Despite all the hardships, we still feel there's good news. While we work through these challenges, we aren't losing sight of the hope and happiness right in front of us on a daily basis. Charlie is not only incredibly strong and resilient, but appears every bit a healthy, bubbly toddler to any casual observer. We're taking every step we can to ensure Charlie's life will be as "normal" as any kid's, without a thought in his mind about cystinosis.

For this reason, we recently traveled over 3,000 miles with Charlie and his big brother, Beckett, to Juneau, Alaska, to visit the boys' grandparents. We've always loved to travel and vowed that we will continue to take our boys on these adventures, regardless of how difficult traveling with a chronically sick child can be. It was a tough decision to embark on such a long trip to a relatively remote place given our new diagnosis and medicine schedule. (As an aside, we thought packing for kids took a lot of stuff before, but that was nothing compared to the small pharmacy we travel with now!) In the end, we made it there and back with only a few setbacks

that were completely and totally outweighed by the amazing memories we made. It was the most special, inspiring trip we could have imagined.

Our optimism is bolstered by the diligent research and development into treatments and a cure for this disease. We also feel so fortunate to have the Cystinosis Research Foundation and the lovely families who immediately welcomed us into their community. They have already spent so much of their own time coaching us through these early days. While we are still adjusting to this life-changing development, we are beyond thankful for the progress that has been made against cystinosis over the last decade, thanks in large part to CRF, and remain hopeful that a cure is within reach. This is only the beginning of our story – Charlie has a condition that we'll have to contend with for a long time, but he remains a happy and chubby kid with no idea anything is wrong. And we intend to keep it that way.

By Shannon Keizer

CALEDONIA, MICHIGAN

power and grace

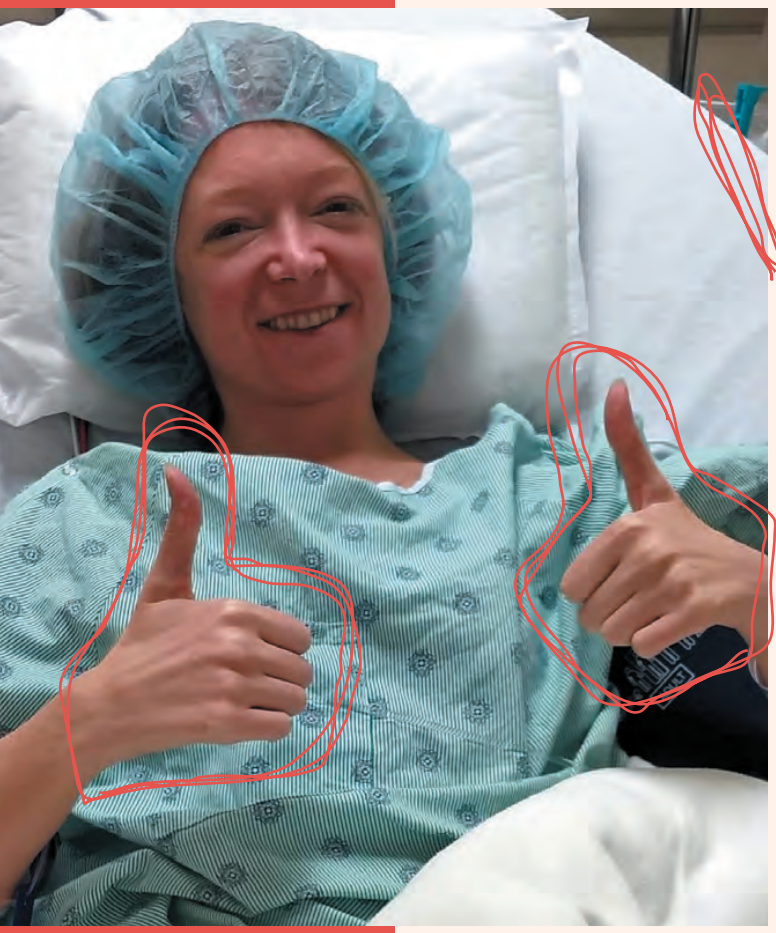
Five years ago if you were to ask what brings me passion, I would have said "travel and adventure." I would have quoted the lyrics of Tim McGraw's "Live Like You Were Dying" and recommended Rocky Mountain climbing, studying abroad in Europe, moving cross-country with only what belongings fit in a Pontiac G6, etc. But now that I've reached the wise, old age of 30, my answer is quite different. Although those things are still important, what I now find most thrilling is making a difference in people's lives. It's about building relationships and being part of something bigger than oneself. It's stepping out of my comfort zone and being bold for the benefit of others.

Last April, I had the opportunity to do just that when I was invited to speak alongside Dr. Stéphanie Cherqui at the CIRM Symposium in San Francisco. Talk about a thrill. If you're familiar with CIRM, you may recall the generous grant awarded to Dr. Cherqui toward the stem cell and gene therapy trials. As part of the grant process, the two of us were tasked with presenting the needs of our community to members of CIRM and the general public. My goal was to answer the question, "How do you, as a patient with cystinosis, feel about the cure?" This has been a recurrent inquiry not only from CIRM but many others in everyday life as well. Here's a snippet of what was shared at the symposium…

Others have said that I seem to handle every curve cystinosis throws at me with power and grace. But being completely real, this entire past year I was a hot mess. Long story short, last October, a CT scan revealed three-inch, coral shaped stones filling both of my kidneys. Typically, people with cystinosis don't get kidney stones, but the rocks I grow could be on display at the Smithsonian.

Little did we know this was the beginning of a long, strenuous journey. Since then I've endured nine hospital stays, including four surgeries (invasively through my back and bladder), various sepsis infections and four months of c-diff. The trauma to my kidneys caused function to tank to about 18%, and I was evaluated for transplant.

Fast-forward to December 25. Christmas was my rock bottom. What was supposed to be the most wonderful time of the year was spent in a pool of tears on the basement floor. In that moment, I imagined all my friends, married with children, opening gifts with their families. Meanwhile, I was just trying to physically make it through the day. I broke down, asking questions like, "Why is it that I do so much right in this world and get crap in return? If SO many people are praying for me, why can't I catch a break?" Life continued while I was operating on autopilot. It was as though I was stuck at the bottom of a pit wondering if there would ever be a way out.



WE ANTICIPATED THIS FOR SO LONG. BUT NEVER FATHOMED IT WOULD HAPPEN IN MY LIFETIME.

That following January while in the hospital, I received a call from Nancy Stack with news that the FDA had approved human trials for the cure. Immediately, overwhelming emotions and excitement came flooding over me. We anticipated this for so long, but never fathomed it would happen in my lifetime. I remember thinking, "The only thing I've ever known is cystinosis. Who/what would I be without it? How would this affect my future?"

Over the next several months, news of the trials sank in, my kidneys began to heal and I gained a new sense of confidence. I made the bold decision to apply to nursing school and officially started classes this fall. I step into the unknown with faith that everything will work out. The cure has given me so much hope. It's given us all hope!

Our stories are already being used to make a difference in the world. And someday, we may be able to say, look where we were. See how we struggled, hit bottom, persevered, and here we are today, free of cystinosis and thriving. Fear for our futures has been replaced with excited anticipation. And we have Stéphanie Cherqui and these stem cell trials to thank for that.

From all of us, Stéphanie, we sincerely say THANK YOU. And we are behind you 100%!

OUR CRF COMMUNITY

After the announcement of the approved clinical trials, I was curious about the reactions of the community. What were others' hopes and dreams? How did they feel about the cure? So, I reached out on social media and asked. Here were a few of the many responses.

- Three-year-old Eleanor wishes not to do eye drops or get pricked with growth hormone each night. She aspires to be a doctor when she grows up.
- Brothers, Samuel and Lars, are tired of swallowing so many pills and would love to make it through the night without wetting the bed. Sam desires to be a dad and a historian, while Lars would like to become an inventor.
- 10-year-old Mary is a rockstar! This future doctor already changes her own g-tube (proud godmother right here). She looks forward to the day when she no longer throws up every morning.
- 15-year-old Tina is in the process of receiving her first kidney transplant. She plans to be a pediatric nephrologist.
- Since Becca's wedding a year and a half ago, she's been hospitalized nine times with 11 infections and will soon receive a second transplant. She dreams of being healthy, finishing her degree and raising a family.
- Newlyweds, Bryan and Alex, met at a cystinosis conference years ago. They aim to start a family and watch their future children grow into adulthood.

As I've gotten to know each of these people over the years, I've watched their ambitions evolve. Life experience has a way of doing that. Just as my passions have matured in the last five years, so have my aspirations.

I'd always liked the idea of being a nurse but never pursued it, because honestly, science classes are scary, and I had to consider practicalities most others don't think about. For instance, if muscle wasting in my hands progresses, will I be able to provide proper patient care, give shots, insert IVs? Can I physically handle being on my feet 12 hours a day? Is it wise to continuously surround myself with contagious illnesses? Is this a realistic career choice?

STRENGTH IN NUMBERS:

A diary of a family coping with cystinosis

By Shannon and Kevin Lawrence, Maddie's parents
LONDON, ONTARIO, CANADA

June 7, 2018: The day our lives changed forever.

For most parents, this is the day their children are born. For us, it was about a year after Maddie's birth — the day she was diagnosed with cystinosis. Despite the many challenges she faces, Maddie is the strongest, most resilient one of us all. She is what motivates us to keep fighting. She will never give up. Neither will we.

Allow us to bring you into our home to show you our "new normal." This is a look into the life of the Lawrence family: mom, Shannon, dad, Kevin, Hudson, age five, and of course, our sweet Maddie.

Midnight — We've just finished washing about 300 syringes. Tomorrow is "medication draw up day." Boy, it is a lot of work. Fortunately, my mom comes over to help. Time to get some sleep.

3 a.m. — Beep, beep, beep. I always need an alarm to wake us up. It's medication time. The house is so quiet. Maddie is sleeping so peacefully, like a little angel. It's been a year, but sometimes I cry when giving these 3 a.m. meds. I used to cry every night. I just wish she didn't need all these medications and supplements.

10 a.m. — Maddie is super-cuddly. This isn't typically like her, but she is getting more affectionate. Her sweet hugs are just what I need to keep fighting.

6 p.m. — It's the night before Maddie's next clinic appointment. We're trying to get Maddie's urine sample. She hates doing it at the clinic. It's going to be a bit harder than usual now. She is in the process of potty training "herself" and the little monkey keeps teasing us. "Pee pee," she tells us with a smile but not going. Like everything, eventually we get it.

10 p.m. — Kids are in bed. It's the night before her clinic. I'm anxious. How will the blood work go? Will it be one poke or two? Will Maddie cry? Will she gain weight? Height? Will she be okay with the doctor? Will we have to increase her meds?

8 a.m. — Off to Maddie's clinic we go. Well, at least we thought. We forgot the urine. We realize it in time. We head back home and get it.

1 p.m. — Yes! The clinic is over. Maddie gained in both weight and height. We are beyond happy! Our hard work is paying off. Because of the weight gain, we need to increase three meds. Increasing meds used to hit me really hard. It made me feel like we weren't taking good care of Maddie. But over the past year, I've been working on not taking it so personally. The increase is due to her growth.

3 p.m. — The kids are playing outside in the sandbox making us "food." It is moments like these where I forget cystinosis and remember some of the ease and innocence of our life pre-diagnosis. Yes, I still think of our lives "pre" and "post" diagnosis.

10 p.m. — Hudson and Maddie have been asleep for a few hours. Hudson just woke up and climbed into Maddie's bed to sleep. They're cuddling. It makes me so happy that they have each other and can provide comfort to each other.

The saying "it takes a village to raise a child" rings true. Thank God for the community of help we have in family, friends, physicians and CRF. We wouldn't be where we are today without our community.

'BLOWN AWAY' BY THE SUPPORT AT OUR BARBECUE FUNDRAISER

When we began planning a fundraiser for the Cystinosis Research Foundation, we never dreamed we would be able to raise more than $20,000. Kevin's brother, Brad, wanted to host our family's first fundraiser for CRF. Brad has lots of experience, as he used to sit as a member on a hospital board responsible for fundraising. We had about 100 people attend our silent auction, with prizes donated from our friends and family. We were blown away by the support.

It was a beautiful day. The kids had an amazing time. There was a bouncy water slide, bouncy castle and balloon artist for the kids. A musician, and childhood friend of Kevin's, performed for us. We raised a little more than $10,000, but a big tech company has pledged to match our donations dollar-for-dollar. It will end up being $20,000 to CRF. The tech company has also selected CRF as one of the beneficiaries for its golf tournament. It will be donating $10,000 to CRF in Maddie's name. We cannot wait to host our next event and get closer to the cure!


Jersey Mikes
SUBS
Date: 5/1/19
HEARTS FOR HADLEY
$ 4510 88
Hearts for Hadley

Day *of* Giving!

By Marcu and Ben Alexander, Hadley's parents

BOISE, IDAHO

Hearts for Hadley was selected as the local charity by the Boise Jersey Mike's Subs restaurants for their "Month of Giving" campaign. This year marked the 9th Annual "Month of Giving" and nationwide raised $7.3 million for charities. The campaign culminated on March 27th with the "Day of Giving" when 1,500 Jersey Mike's restaurants donated 100% of sales, not just profits, to more than 200 different charities across the nation.

Jersey Mike's and our community raised $9,510 for Hearts for Hadley! We had the pleasure of meeting with their employees before the event began and introduced them to Hadley and our family. We had the opportunity to share about cystinosis and provide them with brochures, H4H stickers and t-shirts they proudly wore all month long.

Guests were provided with big, red cut-out hearts when a donatŠn was made, and they could write their name and a message of hope to be hung in the restaurants.

We visited both locations several times throughout March and I was emotional each time I saw the slew of red hearts outlining the restaurant and the words of encouragement that were written on them.

The "Day of Giving" was a huge success! The company where Ben and I work ordered sandwiches for the entire office to help support Hearts for Hadley. We enjoyed both lunch and dinner at Jersey Mike's that day and ran into so many friends and family who were also there to support our cause.

We're so grateful to live in a city that readily bands together to support one another!

trust and understanding

By Erin Little, Olivia's mom
PORT ELGIN,
ONTARIO, CANADA

When Olivia was born, I never imagined she would become a sick kid and I never thought I would be co-parenting with a nephrologist. If you would have asked me eight years ago what a nephrologist even does, I wouldn't have had a clue. I also admit that I didn't know a thing about our kidneys. I knew they were an organ and they looked like a bean I often put into my chili recipe.

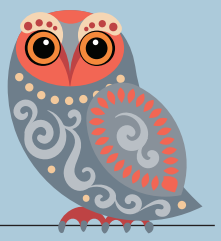
Fast forward to today and I know fancy words such as creatinine, cystatin C and urea. There are still tons I don't know, which is why I am grateful for Olivia's nephrologist Dr. Grimmer. I'll wholeheartedly admit that at first I didn't like her, mostly because I was angry that Olivia had a disease and on top of that she was telling me what I had to do and how to do it. She kept adding different supplements for her Fanconi syndrome, which I didn't understand. She wanted to put her on different medications that I knew nothing about. I never wanted our children to have Tylenol, and now our washroom looks like a mini pharmacy. I didn't like the diet that they recommended for Olivia, and I wasn't budging on feeding her through her g-tube. I probably wasn't the easiest mother to deal with either: I grasped for control over the situation wherever I could—sometimes for the right reasons and other times because I wanted to have a say in how we cared for our daughter.

It has been a long road getting to where we are today and there are some key things I have had to learn during the process. One of those things being trust and that it goes both ways. If I wanted Dr. Grimmer to trust me, I had to trust her. When it comes to Olivia's kidneys, understanding how they function and what her blood work numbers mean, I leave that up to her. She is our kidney goddess. I have had to accept that I don't need to know what every lab reading means and, at the same time, ask lots of questions to help us understand. She also had to trust that I was doing my best at home and doing everything that I could. I leave the kidney knowledge up to her, but I became Olivia's expert until she can do it for herself.

I also had to learn to accept the fact that this woman will be in our lives for the next 10 years, and I needed to let her get to know us and build a healthy relationship. We see her more than we do most of our family. She has watched Olivia go from extremely sick to a beautiful, healthy little girl. Dr. Grimmer is so proud of her growth chart that she at one time wanted to hang it on her own refrigerator. She was there for us when we got pregnant with Harper and we will never forget the day she called to tell us that Harper was healthy. When we go through stressful times as Olivia's advocates, she calls just to check-in on Chad and me. She gives me space to cry and doesn't want to fix it; she just

ALTHOUGH IT HAS TAKEN ME SOME TIME, DR. JOANNE GRIMMER IS THE BEST CO-PARENT I COULD HAVE EVER ASKED FOR.

READ ABOUT THE SWING, SHOOT AND LIV GOLF CLASSIC ON PAGE 69

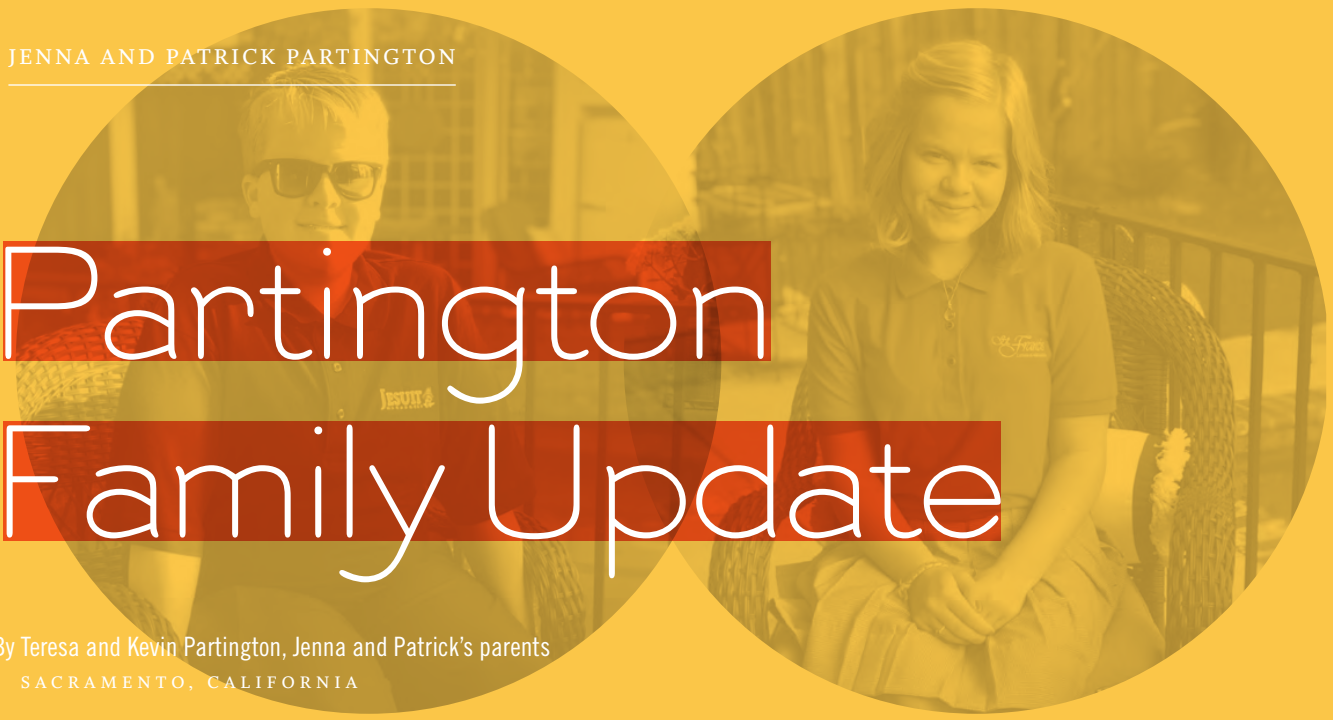


lets me get it out. Had I not accepted her into our lives, I don't think Olivia would be doing as well as she is. She is the first person to remind me that she has the easy job of reading labs and writing prescriptions and we have the hard work of caring for Olivia.

Lastly, I think the most important piece of our relationship is appreciation for one another. We are so blessed to have this amazing woman show up every three months to care for Olivia. I am so blessed to be able to be home with our girls, to raise them and educate them.

There are so many different ways to care for and appreciate our partners. We love taking Dr. Grimmer and our team special cookies. Dr. Grimmer once asked if she could have our chocolate chip cookie recipe and the answer was "no" because then she could just make them whenever she wanted. She can get the recipe when Olivia is cured, and she is no longer her patient.

Dr. Grimmer has been with us from day one and although it has taken me some time, she is the best co-parent I could have ever asked for. I do an amazing job at giving Olivia what she needs and it's easy because I know I have Dr. Grimmer on our side doing her best for Olivia. I never thought this was going to be my life, our life, but here we are doing our best and making the most out of it. Dr. Grimmer has been one of the greatest gifts on this journey. Her support, encouragement and reassurance that we are doing an amazing job as Olivia's parents gives us strength to keep working hard, to keep caring—and the strength to get through the next stage.



Partington Family Update

By Teresa and Kevin Partington, Jenna and Patrick's parents
SACRAMENTO, CALIFORNIA

Greetings! Autumn has arrived, and Jenna and Patrick are getting settled into a high school routine. To say it's a learning curve for all four of us is an understatement!

The months leading up to high school were busy as we tended to the cystinosis-related medical needs of Jenna and Patrick (trying to accomplish as much as possible during summertime days off). In July, Patrick had guided growth implants surgically removed from six locations on his legs and ankles. While an infection set him back a couple of weeks, his recovery was swift. The procedure worked for Patrick; his genu valgum (knocked-knees) straightened significantly, and we hope they remain stable as he continues to grow.

In mid-July, Patrick and Jenna were seen at Stanford University's Lucile Packard Children's Hospital for their kidney transplant evaluations. It was a full day appointment, including visits with doctors of nephrology, psychology, nutrition and a pediatric transplant surgeon. Jenna and Patrick have about 30% kidney function, which makes them candidates for the donor kidney transplant list. It is predicted that Jenna and Patrick will both receive a kidney transplant during their high school years. We have dreaded this news but know that a kidney transplant will much improve the kids' overall mental and physical stamina. It will also make them candidates for the stem cell and gene therapy trial in a few years, when pediatric patients may enroll. We learned that Kevin is a living-donor match for both kids and that my breast cancer history rules me out as a donor candidate. As the time for transplant nears, Kevin will be more thoroughly evaluated as a candidate for whichever of the twins needs one of his kidneys first. If this seems altogether overwhelming, unnerving, rare and scary, it's because it is. I'm impressed and relieved by what Jenna and Patrick have lived with and overcome in the past 15 years. I remain hopeful, yet have a difficult time contemplating what the next 15 years might bring.

Our family and friends (YOU!) have been dedicated supporters of Jenna & Patrick's Foundation of Hope since 2006. Our HOPE comes from progress made in the research and treatment of cystinosis, thanks to the Cystinosis Research Foundation.

Here is a summary of the JPFH fundraisers that have taken place in 2019:

- Our niece Lindsey Partington and her new husband Jack made donations to JPFH a wedding gift request, raising over $1,000 in honor of their nuptials. Quite a charitable start to their lives together!
- We thank our friend Michael Heller for organizing and raising $30,000 at "ICE FEST 2019." The first-ever ping pong tournament at the Ice Blocks development in downtown Sacramento was a massive hit with Craig Zarro taking home the championship trophy!
- Thank you to our dear friends Shannon Bell of NorCal Beverage and Uncle Tommy Walcott of Colliers International, for making JPFH your charity of choice for Sacramento's Capital Cup Charity Golf event, to take place September 27-29, 2019.
- Finally, The Annual "Swing" Golf event to benefit JPFH will take place on October 3, 2019, at Catta Verdera Country Club in Lincoln.

We will once again present the proceeds from these events to the Cystinosis Research Foundation at the Natalie's Wish Gala on April 18, 2020. While asking for dollars is never easy, we remain dedicated to the effort of raising funds for CRF, knowing that every dollar is put to work swiftly and thoughtfully.

READ ABOUT THE CAPITAL CUP GOLF TOURNAMENT ON PAGE 75

We've made dear friends in our years as members of the cystinosis community and many of them have been on our minds. Prayers are with our pal Tina Flerchinger, from Clarkston Washington, as she prepares to receive a kidney from her father in October. Tina is a year older than the twins and she has always paved the way. She took the medication Procysbi® first, then started Cystaran™ eyedrops. Tina taught the kids how to do a growth hormone injection over a video-chat session. Tina went to high school first, providing us a sneak peek into a challenging life event made doubly challenging by chronic illness. She is a dedicated pen pal, a faith-filled young lady with a sweet disposition. We are rooting for Tina to feel better soon.

Our friend Hadley Alexander (page 30) recently received a diagnosis of osteochondritis dissecans or OCD (a joint disorder in which cracks form in cartilage). OCD is the painful condition that ushered Jenna into her ongoing plight with orthopedic complications. The fact that Jenna and Hadley (who look remarkably alike) are dealing with this unique diagnosis is proof enough to me that OCD is related to cystinosis. We are grateful that Dr. Kathleen Rickert, a pediatric orthopedic surgeon at Rady Children's Hospital of San Diego, has recently arrived on the scene as a clinician for cystinosis patients. Dr. Rickert is holding cystinosis clinics, gathering data and looking into the root cause of conditions like OCD, genu valgum and osteopenia, all common in cystinosis.

Last week, somewhere on the other side of the world, a 10-year-old boy died from complications related to cystinosis. He lived in India. He had very little access to medical care, no life-preserving medication, no family support events or cystinosis clinics to attend. Umesh, his father, posted a photo of the child on Facebook; he was a darling, tiny boy with the subtly familiar facial characteristics of a person with cystinosis. In the photo, he sits with his legs folded in and crossed over one another (the very way Patrick prefers to sit). The contrasts in the access to care and quality of life that Jenna and Patrick enjoy compared to the boy in India are not lost on us. Our deepest sympathies are with this child's family.

We are humbled by the strength and support of those around us. We cling to faith, hope and gratitude, doing all we can to help Jenna and Patrick happily persevere as they grow into adults who will navigate life with cystinosis themselves.

Jenna & Patrick's FOUNDATION OF HOPE

SWING GOLF EVENT

Jenna and Patrick's Foundation of Hope had a successful 12th Annual "Swing" golf event on October 3, 2019. A full field of golfers enjoyed the beautiful day at Catta Verdera Golf Club in Lincoln, California. Thank you to our title sponsors: Gallelli Real Estate, GPR Ventures and Cushman & Wakefield. Congrats to David Lichtman and his winning foursome! More than $150,000 was raised—which will be presented to CRF at the Natalie's Wish event on Saturday, April 18, 2020. Thank you to the generous sponsors and longtime supporters and the Sacramento community for their commitment to cystinosis research and the cure!

TEENAGER STATUS

By Brian Sturgis, Henry's dad
SANDPOINT, IDAHO

Summer started with warm weather and a trip to Clarkston, Washington, for Henry to spend five days with Tina Flerchinger. The trip included a bonus, because Sam and Lars Jenkins from Salt Lake City stopped by for a couple of days! They all had a great time catching up and hanging out! Henry really loves to spend time with his close friends that he has met because of cystinosis. After that, it was summer on the lake spending time with friends and family. In July, Henry officially became a teenager, the big 13! We finished up the summer with a weekend camping trip to Priest Lake, a trip that we haven't had the opportunity to do the last couple of years. It was great to do it again!

September marks a big transition: the transition to 7th grade and middle school! The weeks leading up to the start of school were met with anxiety and uncertainty for Henry. Transitioning to middle school is a big deal for all kids, but you throw in cystinosis and it adds another level of complexity. It makes me think back to when I started 7th grade. I was nervous too – new school, new kids, "Will I know anyone in my classes?" "Is homework really as much as they say it is?"...and, no more recess! Henry has all these same worries, plus: his medicine schedule, side effects from meds, will he feel good, can he keep up...?

As parents, gone are all the people from the elementary school that have been advocating for Henry for the last six years. Educating teachers and counselors about Henry's needs starts all over again. We thought we had a solid plan in place. We had met with both elementary and middle school teams. We felt reassured that all IEP information and Henry's needs were being passed on to the next team. But the best laid plans don't always work out. When we attended the 7th grade orientation, Henry's class schedule was completely incorrect, and it was clear we had more work to do. We met with his IEP case manager and were reassured it would get sorted out, but with only one day before the start of school, it created some anxiety.

The first day of school arrived and off we went. Henry was able to find all of his classes (albeit the wrong ones); he learned he had enough time in-between classes and that lunch could still actually be considered recess. We met with the school counselor and Henry's IEP case manager and had a new schedule in place by the end of the first day of school. Henry was able to get some of the classes that he really wanted and is happy! The next day went better and the transition seems to be going better, too.

As we roll into the start of another school year, we also start thinking about our next fundraiser, 2,400 Feet of Schweitzer (March 28, 2020). The pre-planning starts now, and we will be knee-deep into the planning in another month or two.

Our entire family continues to wait patiently for the results of the current stem cell trial, praying that it works just as well in humans! I'm asked frequently if I know any more about it. The two new potential eyedrop delivery systems are also talked about a lot; Henry really wants to be on those. He hates the eyedrops.


ID 1831 X

BROOKE'S JOURNEY
FROM PRE-K TO KINDERGARTEN!

This past June, my daughter Brooke completed her first year of public school – full-day Pre-K 4. In hindsight, the beginning was a lot more difficult for me than it was for her. Giving up control and trusting virtual strangers to administer meds and care for my daughter for six hours per day, five days a week was a lot, especially when she has been home with me full-time since she was born. The first few weeks, I probably reached out to Brooke's teacher and nurse daily. To my surprise, Brooke quickly adapted to these changes and the new environment, and I began to relax and develop a sense of security and comfort. She came out of her shell beautifully. It's amazing how much we can learn by watching our children grow!

What made it easier for my husband Clay and I was how much her teacher, nurse, therapists and paraprofessionals loved her, looked out for her and communicated anything they believed may be noteworthy. Every day at parent pickup, they would inform me if she ate a lot of lunch, if she didn't drink enough water and even if she napped at rest time (a noteworthy occurrence for Brooke!). I always received regular calls, emails and updates from her nurse, therapists and teacher. Brooke's teacher would even randomly send me messages to let me know that Brooke was having a great day! I truly believe that by sitting down with Brooke's team multiple times prior to school starting, giving them a plethora of cystinosis information and explaining everything and anything that may occur, we alerted them to the importance of cystinosis and our desire to have open lines of regular communication and involvement. It may have been overkill, but it gave us the comfort we needed as we began navigating Brooke's school experience. Her team was very open to this and helped us adjust to Brooke's development into an independent school age child; we were pleasantly surprised by how smoothly Brooke's Individualized Education Plan (IEP) functioned and how focused her team was on her success.

Brooke began kindergarten this September, and while she has retained the same physical therapist, she has a new teacher, paraprofessionals, nurse and case manager. Even though this means there will be a learning curve (and they'll have to get used to

READ JILL EMERSON'S ADVICE FOR NEW CYSTINOSIS PARENTS ON PAGE 44

my regular check-ins!), I wholeheartedly have faith in the IEP process and our school district's dedication to Brooke's education and success. While we cannot imagine a more perfect IEP team than her Pre-K team, we know that she has been paired with an amazing kindergarten team and we feel so fortunate and reassured that Brooke will continue to have a wonderful elementary school experience.

We had so many proud moments during the year: when Brooke learned to write her name, when she took her classmate who got hurt on the playground to the nurse and promised him that the nurse "isn't scary at all," and when she decided for Career Day that she wanted to be a school nurse. Most of all, we are so proud that Brooke adjusted so quickly and seamlessly and had an amazing Pre-K school year. As she continues kindergarten, we are excited to continue to see her grow and develop into the independent, funny, smart and empathetic girl she is.

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

Brooke

Brooke and her teacher, Kim Russomanno.

SOPHIE BETOURNAY

READ ABOUT THE HOUSE OF LUCK FUNDRAISER ON PAGE 74

Finding Victory

A MOTHER'S MESSAGE OF HOPE

By Nicole Chatelain, Sophie's mom
KANATA, ONTARIO, CANADA

When I was 20, I got my second tattoo – a small, intricately drawn "V" on my lower left calf. And as is often the case with tattoos, people like to ask me what it means. My name doesn't begin with a V, and I don't have any close family members whose names begin with V. It usually throws people for a loop.

The answer is simple: my name means "victory." I've always liked that my name has such positive connotations, and that's the whole basis behind the tattoo—it didn't really occur to me how exactly I should define victory or how I would strive for victory in my life. I was just a college kid getting a cool tattoo.

This all happened before I ever met my husband, before we started a family, and long before I had ever contemplated life as a rare disease parent. 15 years later, I'm the mother of two remarkable children, including one with cystinosis. And, I could certainly write a lot about how drastically this disease has changed our lives over the past four years, or what receiving a diagnosis was like. But if you'll forgive me, I don't want to write about those things. If you're reading this magazine, you might have lived these same experiences yourself. You might know all about the fear, the anger, the exhaustion and the heart-wrenching sadness that can swallow you up during the early days of a cystinosis diagnosis. These wounds are still raw, and I'm not yet ready to revisit them.

What I'd so much rather tell you about is my daughter, Sophie. She's a delightful little girl who is curious, funny and strong. Sophie loves bubbles, jigsaw puzzles, building blanket forts with her dad and playing outside on a crisp fall day. She loves to laugh and to learn. She loves her little brother, Xavier, and always tries to teach him new words—she's just as thrilled as my husband and I when she hears him say something new for the first time. Sophie is a deeply profound and wonderfully unique person, who's given us so much more joy than cystinosis has ever given us pain.

I've felt more purpose and sense of accomplishment in the four years I've been a mother than I did in all the 31 years I lived before then. My family gets me through the hard days, of which there are certainly many, and they're there to share in the good ones—of which there are, thankfully, far more. Cystinosis has not stolen our daughter from us. Rather, we're seeing more and more every day the ways that Sophie is starting to steal herself back from the disease.

I hope for so much for my children. I hope they'll be happy, I hope they'll be healthy, I hope they'll always know and feel love. And, if they do, I'll know that I've found that victory I was looking for with my second tattoo 15 years ago. Many people with tattoos will tell you how the meanings they give to their various pieces of body art can shift and change over the years. Even if I didn't know then what victory meant to me, I know now. I know what it means to fear, and to hope, and eventually, to believe – to believe that change is possible, and that it's coming. And I do believe that a cure is coming for us. It's not a wish or a dream, but an inevitability. We will get there. This powerful community of rare disease warriors will get there together.

One day, I will add a date to my "victory" tattoo. It will be the day that Sophie is cured of cystinosis. And that day will be a day of victory for us all.

SAMANTHA "*Walnut*" GROVER

10/5/1988 — 4/24/2019

Sam had a true joy for life. "Every breath is a second chance" and "just keep swimming" were two of her tattoos. A daily reminder to not take life for granted and to keep going, even when times are tough. Times were tough, and times were great. She faced each obstacle with more positivity and grace than I thought possible. She was no saint. She had days where she was crabby, dramatic, selfish, childish, needy, very anxious and frustrating. However, we all have those days. She deserved more of those days, but hated confrontation, being a burden, and truly just wanted everyone to get along. She would plaster a smile on her face whether she felt like it or not, and just kept swimming.

By Karen Grover, Samantha's aunt
EXETER, NEW HAMPSHIRE

Samantha had a big heart. She would cry with joy when watching Ellen DeGeneres giving gifts to those in need. She loved to secretly pay for lunch for a person eating alone when we would go out to eat. She adored the NHSPCA fundraiser and all animals, especially her Yorkie, Bella. Her eyes would fill with tears of delight when she would remember winning an auction item, and then donating it back to raise more money. She dreamed of going to every door in town and asking every person for just 10 cents for CRF. "Every little bit counts," she would say. "Even if it is only a dime, imagine how much money we could raise; everyone has a dime to spare."

Sam gained a very supportive relationship with family and friends in the last several years of her life. She became best friends with her little sister, and hung out with her brothers and sister-in-law. She loved nothing more than hugs from her daddy, who called her "Walnut" and girl time with her step-mother. She thrived on the time spent with her girlfriends, and Sam loved living next to her cousin "Zachaarrry." They were more than cousins, more like siblings and maybe too much alike.

She regained time missed with her aunt and uncle and loved being called "Spammy" again. She lit up like a Christmas tree when she would spend time with her niece Leah, and personally I believe this relationship helped carry her through her most challenging days. She mended the relationship she had destroyed with her mother and that lifted a weight off her soul. After all, every girl needs her mom and every mom needs her little girl.

CRF opened a new world to Samantha. I will be forever grateful that she connected with them in the last few years. She learned so much about her disease, herself and finally met people who could truly understand her, "her people." Her life was completely and positively forever changed. She made friends that meant more to her than they will ever know. She was ready to give up at one point, but I reminded her of her promise that she would make it to California for the conference. Well, she did that. Then, in true princess style, she passed in Beverly Hills. Sam is a hero to me—but not by the manner in which she died, but in the way she lived her life.

LYNDSEY BEELER

1/19/1980 — 10/4/2019

By Deanna Beeler

Lyndsey Beeler, a cystinosis patient pioneer and all-around delightful person, died peacefully in her sleep on October 4. Her parents, Debbie and Alan Beeler, were at her side, as they had been constantly since she returned from the hospital under hospice care. She was 39 years old.

Tiny Lyndsey was born on January 19, 1980, with a thyroid tumor that kept her from breathing. Before she could even meet her parents, a nurse grabbed her up and ran down the hall to get her stabilized; then she was rushed to the Oregon Health and Sciences University hospital for emergency surgery to remove the tumor.

That surgery was a success, and Lyndsey was returned home for a couple of years of health and joy. She bounced around light-footed and happy; her big sister Jenée spoiled her, and fondly called her "Bean." She loved the color yellow ("lellow"). Her grandma Evelyn (among others) made sure she had plenty of "lellow" clothes and toys. She was a ray of sunshine. Everyone loved her.

At the age of 2, she was diagnosed with cystine cystinosis, a rare genetic disorder for which there was at that time no cure or even serious treatment. Lyndsey drank a lot of water, in the hope of dissolving the intracellular crystals caused by the cystinosis, and ingested electrolytes to replace those washed out by the water. Many family members had themselves tested to see if they were good donors, and many of them traveled to La Jolla to take part in Doris Trauner's study. For a long time, the science seemed to stay ahead of Lyndsey's body's deterioration, and in the mid-eighties, she began to take cysteamine, a disgusting bitter substance that slowed the disease's progress. Lyndsey and her parents learned as much as they could about the condition. They read, went to conferences, and stayed in touch with researchers. They had been told to expect her not to live past 10 or 12, and her family marveled when she made it to her first kidney transplant at 13, beating the odds, and then her second at 20. At each milestone, she smirked to friends that when she was first diagnosed the doctors didn't think she would make it to 10, and now look at her! She sure fooled them!

It was hard. So many doctor appointments, blood tests, the two kidney transplants, and all the poking and prodding — but throughout it all, she maintained a cheerful outlook and zest for life. She crammed many intensely wonderful times in her short life. Her many relatives and friends knew their time with her was limited and made the most of it, and Lyndsey was up for it all! Astronomy, hiking and photography with her dad, art, quilting, jewelry and candle making with her mom, going out dancing with her fiercely protective big sister, having rowdy discussions with her family and friends. One aunt worked with her on genealogy, and Lyndsey triumphantly traced her ancestry back to the Revolutionary War and became a member of D.A.R! Even when she was in pain and had lost much of her hearing, she hugged her family tightly.

When this recent rough patch began, her family hoped she would work her old magic and prove the doctors wrong again. But no such luck.

It was easier for her family to care for her than to go through with Lyndsey's hospice decision. But she knew. A month before her death, during one of many hospital stays, she appeared especially concerned about worrying her parents. Pressed to explain what was troubling her, she said: "I think I'm dying."

Her family cannot begin to express their grief and the immensity of Lyndsey's loss.

FOLK POEM *by Jenée, Lyndsey's sister*

As I sit in heaven and watch you every day
I try to let you know with signs I never went away
I hear you when you're laughing and watch you as you sleep
I hold my arms around you to calm you as you weep.
I see you wish the days away begging to have me home
So I send you signs to show you that you are not alone
Don't feel guilty that you have life that was denied to me.
Heaven is truly beautiful, just you wait and see.
So live your life, enjoy yourself, laugh again, be free
And know with every breath you take you're taking one for me.

WHAT I KNOW NOW, THAT I WISH I KNEW THEN:

ADVICE FOR NEW CYSTINOSIS PARENTS

By Jill Emerson, Brooke's mom

HAMMONTON, NEW JERSEY

My daughter Brooke was diagnosed at 16 months. She is now five and started kindergarten this September. When I talk to parents of newly diagnosed children, they often ask me questions or raise concerns that I also had and felt when I was in their shoes. I, too, had fears and concerns similar to theirs, and when I tell them how things are for our family now, four years in, I often think, "Wow! How far we've come, and how much I wish I knew then what I do now!"

THERE WILL BE PEAKS AND VALLEYS, BUT IT DOES GET EASIER!

The beginning is tough. It is lonely, it is scary, and more than anything, it is overwhelming. There will be moments where you feel defeated or feel like you don't know how you'll survive another day on such little sleep.

It will get easier and you will get better at it! I wish I knew then that I would look back at the stains on my kitchen ceiling from a Procysbi® explosion or on the roof of my car from a bolus feed mishap, and they would now make me chuckle and remember how tough it used to be and how far we've come. I wish I knew then how adept I would get at mixing and administering meds in a public place, without caring who sees or what people must think. I wish I knew then that the little things don't matter, and that I can handle the responsibility of cystinosis better than I ever imagined.

IT IS OK TO ASK FOR HELP!

Help can come in many forms. It can be help from other cystinosis caregivers, friends or family members, or a trained therapist. Help can also come in the form of a yoga class, or a book club, or anything that helps you take a breath and can ground you. In the beginning of our journey, my husband Clay and I thought we had to do it all. However, as cystinosis became my identity, I lost a little bit of myself.

I wish I knew then that it was OK to admit that I couldn't do it all. I wish I knew then that trusting others to step up and do meds or watch Brooke was safe, and even healthy, for both Brooke and me. I wish I knew then that it was OK to vent about the stress of a rare disease, navigating insurance issues and juggling a million medical things at once. I wish I knew then that asking for help would make a world of difference!

NOT EVERYONE FROM YOUR "BEFORE CYSTINOSIS LIFE" WILL GET IT.

You will inevitably categorize some things as "before cystinosis" and "after cystinosis," and that includes friendships and relationships. There may be "before cystinosis" friends or family members who come around, call or include you less often because they don't know what to say, or they don't know why you cancel so often.

I wish I knew then that while I may lose some relationships, I would gain more. I wish I knew then that the silver lining of cystinosis is that I would continually meet people from different backgrounds, states and countries who would become my closest confidants, and who would understand me more than many of my "before cystinosis" friends. I wish I knew that my "after cystinosis" friends would become some of my best friends, and people who I will forever be bound to!

TAKE CARE OF YOURSELF; IT WILL CATCH UP TO YOU IF YOU DON'T!

I remember thinking that I could survive on such little sleep. And I drank a lot of coffee to compensate for the broken sleep to wake up for night meds and feeds. A lot of coffee. I honestly ran on adrenaline, and did well for a few years. I thought that trips to the grocery store alone were "me time," and that was enough.

I wish I knew then that giving myself a break and finding an outlet for self-care were almost as important as the rest of my job. I wish I knew then that heading to the grocery store does not qualify as "me time" and self-care. I wish I knew then that we all need "me time" and by taking care of ourselves we become more present, re-energized, and better parents!

YOUR CHILD CAN AND WILL BE ABLE TO LIVE A "NORMAL" LIFE. IF YOU LET THEM.

This was a hard one for me. Upon diagnosis, some of the first thoughts I had were, "Will Brooke ever go to college? Or get married? Or even play soccer?" I wondered if she would just have the chance at a "normal" life. It wasn't until Brooke reached school age and began living "normally," going to school, making friends and having play dates, and being independent of me and my husband, that I realized that she is completely capable of living a normal five-year-old life, as long as we treat her like a normal five-year-old.

I wish I knew then that by letting go and allowing my daughter to be a normal kid, she would have a normal existence. I wish I knew then that she will find her own path, her own passions and she is absolutely capable of accomplishing whatever she desires. I wish I knew then that even if Brooke was born without cystinosis, she might not go to college, or get married or even play soccer. Truth be told, Brooke probably would not have played soccer like me; she is a dancer and takes after her dad in her artistic and musical abilities, and that is absolutely normal.

IT IS ABSOLUTELY OK TO HAVE "HOPE." DON'T LET ANYONE TELL YOU OTHERWISE.

This is the big one. There will be times where you feel overwhelmed and mourn the life you wished that your child and your family had. You will be scared that advancements cannot come soon enough. It is absolutely normal to feel this way, but I am here to tell you that there is HOPE.

I wish I knew then how close of a reality the cure for cystinosis is, and how thanks to CRF accelerating the course of research into improved treatments, things have dramatically and quickly improved already. I wish I knew how much support and love we would receive each and every day from CRF, Nancy and Jeff Stack, and all of those within the community whom we have relied upon these past four years. And I wish I knew how in our daughter's lifetime, cystinosis will only be a memory.

I wish I knew then that we didn't have to be as scared as we were upon diagnosis, because HOPE was in the very near future. And my wish for all of the newly diagnosed parents is that they know that HOPE is their present.

SOME INSIGHT ON SIBLING SUPPORT

By Lauren Hartz, Landon and Jordan's mom
PITTSBURGH, PENNSYLVANIA

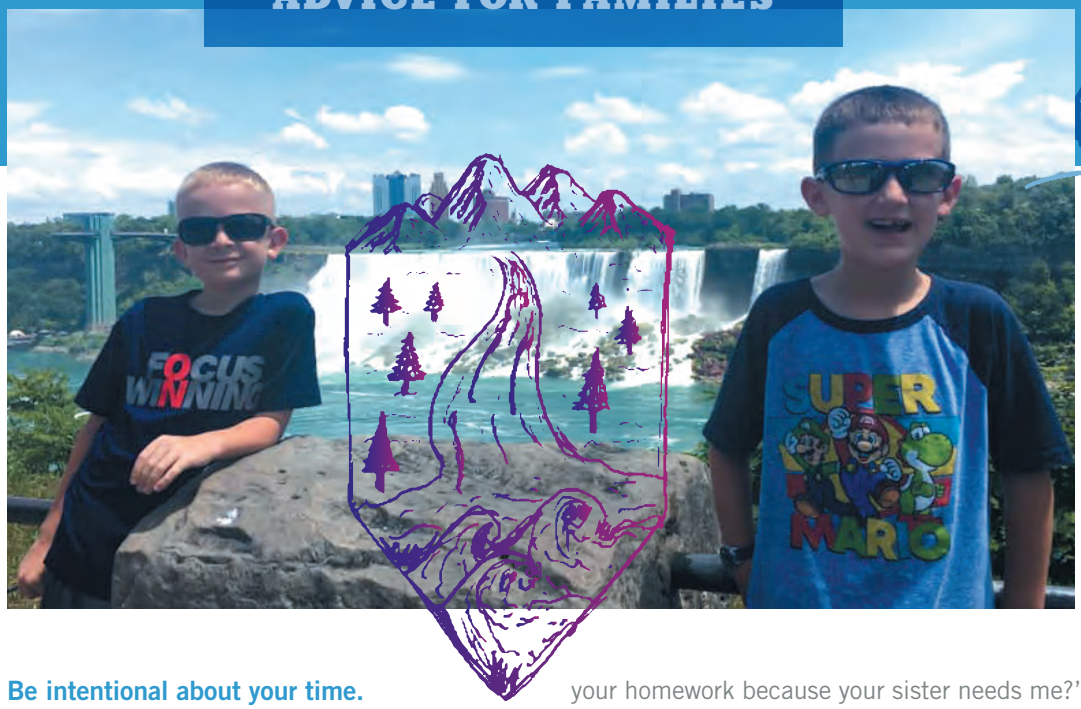
On May 31, 2011, I received a phone call with admission instructions for my son, Landon. I knew that he was to be admitted to Children's Hospital in Pittsburgh for an intensive feeding evaluation. He had stopped eating or drinking anything but water, and when he did guzzle water from his sippy cup, he would throw it back up immediately. "There is nothing medically wrong with Landon," I was told. We were going to be admitted so they could figure out why he didn't want to eat.

I am sure anyone who is reading this article knows where I am going with this. There was something "medically wrong." My boy was diagnosed with cystinosis on June 7, 2011, and didn't leave the hospital until June 27. As I type these words, I am pretty certain that this is the first time that I am communicating this to anyone. But, after Landon's diagnosis was confirmed, I remember standing in the bathroom as his grandparents walked him around the hallway of the hospital floor, and I prayed that I was pregnant. I knew it was unlikely that I was, but I believed at the time that I could never have the second child I hoped for and that if I were pregnant, it would have been prior to my knowing that my child was at risk for having a disease.

My desire to have a second child never went away even when I told myself that it would be cruel to do so. How could I do this to a child knowingly? How could I take any focus and energy away from Landon when he needed so much care? But then, over time, as Landon grew and became healthier and we developed a new norm, my husband Jimmy and I had the conversation again, and decided that we wanted to try to have another child even if there were a chance that this baby would have cystinosis too. The conversation happened in April 2012 and Jordan was born January 15, 2013.

I remember the phone call from the doctor when Jordan was two months old. We were waiting to see if he also had cystinosis. The voice on the other end told me that he did not, and I cried. I cried because I was so thankful, and I cried because Landon's opportunity to have someone in his life who understood what he was going through wasn't going to happen.

The other hat I wear, aside from mom, is mental health therapist. I spent the first part of my career working with children and their families. More recently I started a private practice in which I see some kids and mostly adults. This work has taught me that there are very rarely clear answers and not everything works for everyone. But I hope to offer some insight about how to support siblings of children who have some extra, unique needs such as those with cystinosis.



1. Be intentional about your time. Set aside one-on-one time with your child. It could include a weekly or bi-weekly Friday night date to play mini golf or get ice cream. It also could be a nightly 10 minutes of reading time together, or a short walk before the sun goes down. It doesn't have to be a grand gesture, but something that is just for him/her.

2. Be honest, and appropriate. Your child may have questions and it's important to answer them honestly, but in a developmentally appropriate way. It is okay to tell your child that you have to give it some thought but will get back to him/her with an answer, and be sure to do just that. If this is hard to do, you can reach out to a professional (mental health therapist, pediatrician, etc.) for extra support.

3. Allow space for thoughts and feelings. Whatever feelings your child has are just fine and it's important to communicate that. You can say things like: "I noticed that you rolled your eyes when I gave your brother his medicine. What are you thinking right now?" Or, "is it hard when I have to stop helping you with your homework because your sister needs me?" Try not to react and sit quietly, listen and offer a hug so that your child knows that it's okay to have a hard time sometimes.

4. Identify a support system. You can use your support system in lots of different ways. I sometimes find myself trying to take on too much because I don't want to ask for help. Your neighbor may be happy to hang out with your child with cystinosis who may be too tired to go to her sibling's soccer practice after a full day of school. Your sister may enjoy taking your child overnight once a month for some one-on-one time. Your best friend may be happy to learn how to give meds so that you can go to Cub Scout camp overnight with your child. You don't know if you don't ask.

5. Remind your child that you do see him/her and express gratitude. You may say, "It has to be so hard for you to be quiet after school because your sister needs to rest. I appreciate how thoughtful you are being" or "It was such a long day at the doctor appointment. Thank you for being so patient."

Again, there are no simple answers and you know your child and your child's strengths and challenges. I think the best thing we can do is take care of ourselves so that we can be aware and present with our kids. When we can do this, it is much easier to have conversations and to respond to situations. My self-care involves things like 10 minutes of meditation before I go to bed at night, a warm bath with bubbles after the kids are in bed, reading or listening to a book or writing in my journal. When I am doing these things, I am more content, relaxed and can connect with my family so much easier than when I tell myself that I am too busy to do any of it.

Endocrine Complications of Cystinosis

By Stephen Jenkins, MD, Sam and Lars' dad
SALT LAKE CITY, UTAH

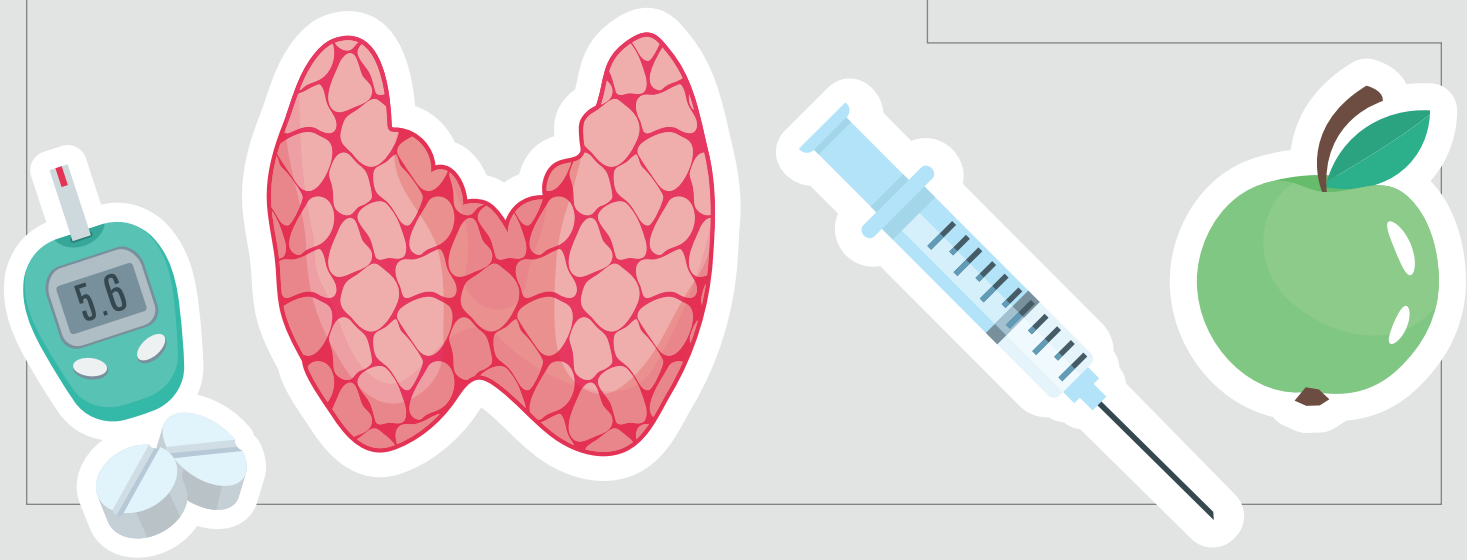
Cystinosis is a genetic disease, which means it affects every cell in the body. The major complications include Fanconi syndrome and renal failure, as well as corneal crystals and muscle disease. Cystinosis also affects many other organs, including the thyroid, pancreas and gonads.

Thyroid disease is the most common endocrine complication in cystinosis and affects about 50% of untreated children by 5-10 years of age.[1] The thyroid is a gland located in the front of the neck, below the Adam's apple. It is regulated by the pituitary gland (in the brain) and secretes thyroid hormones (T3 and T4), which affect metabolism, appetite, nutrient absorption, gut motility, heart rate, heart contractility, oxygen consumption and brain development. Thyroid hormones affect almost every other part of the body. In cystinosis, the thyroid gland is damaged by accumulation of cystine crystals leading to fibrosis (scarring). There is also increased cell death and impaired thyroid hormone synthesis. Treatment with cysteamine, such as Cystagon® and Procysbi®, prevents development of hypothyroidism in the majority of patients.

When the thyroid is sufficiently damaged, it is not able to secrete enough thyroid hormone to regulate its normal activities. Symptoms of low thyroid hormone include fatigue, cold intolerance, weight gain, constipation, dry skin, muscle aches, high cholesterol and irregular periods. The pituitary gland responds by secreting thyroid-stimulating hormone (TSH), which can be measured in the bloodstream. If TSH is elevated in the blood, then the thyroid is not secreting enough thyroid hormone. Fortunately, thyroid hormone can be supplemented or replaced with synthetic thyroid hormone, also called levothyroxine.

In the later stages of cystinosis, the pancreas can also be affected. The pancreas is an important gland in the gastrointestinal system and secretes enzymes to help break down fats and proteins after a meal. The pancreas also regulates blood sugar levels by secreting hormones like insulin and glucagon. When the pancreas is damaged in cystinosis, it is unable to secrete enough insulin, leading to diabetes mellitus. This complication usually develops during adolescence or adulthood and affects 30% of patients within 10 years after kidney transplantation.[2] This is likely due, in part, to the use of steroids and calcineurin inhibitors for immunosuppression.

Symptoms of diabetes include excessive thirst and urination (due to sugar wasting in the urine) and weight loss, and can progress to a life-threatening condition called diabetic ketoacidosis. Diabetes can be diagnosed with an elevated fasting blood glucose, an abnormal glucose tolerance test, or by a lab test called hemoglobin A1c. Diabetes due to cystinosis is treated with insulin replacement. Insulin is administered as a subcutaneous injection. There are long-acting forms of insulin, such as glargine, which are usually given once a day, and short-acting forms such as aspart or



lispro, that are given with meals. Insulin use requires close monitoring of blood sugar levels to prevent low blood sugars (hypoglycemia). Poorly controlled diabetes leads to small vessel blood disease, which can cause blindness (retinopathy), nerve damage (neuropathy), kidney failure (diabetic nephropathy), heart attacks and strokes.

Another important endocrine organ affected by cystinosis is the gonads. Gonadal function and sexual development are usually normal in females, and multiple women with cystinosis have had successful pregnancies and births. The majority of males, however, will experience delayed or incomplete puberty, decreased testes size and low testosterone levels.[3] Testosterone is important for puberty and muscle and bone development. Testosterone can be supplemented with gels and injections.

All males with cystinosis have azoospermia, which means there are no sperm in the semen, and this leads to infertility. However, biopsy of the testes in a male with cystinosis does show intact sperm production, with sperm present in the epididymis. There has been at least one report of a male patient fathering twins through percutaneous epididymal sperm aspiration (PESA) followed by intracytoplasmatic sperm injection (ICSI).[4] This raises questions about the possibility of sperm cryopreservation in young male patients, before significant fibrosis occurs.

As with all complications of cystinosis, the most effective way to prevent disease is to be compliant with oral cysteamine therapy. Unfortunately, even with strict adherence many patients will develop some endocrine complications, so it is important to be aware of the possibilities so that diagnosis of complications like hypothyroidism, diabetes and hypogonadism is not delayed and can be managed appropriately. There is hope that gene therapy, such as a genetically modified autologous stem cell transplant, will be effective for treating and preventing endocrine complications in cystinosis.[5]

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cystinosis RESEARCH FOUNDATION | TOGETHER WE SHINE BRIGHT

NEW MEDICAL AND SCIENTIFIC ADVISORY BOARD MEMBER

Dr. Barshop is a certified Clinical Biochemical Geneticist and has cared for patients with all sorts of metabolic disorders for more than 30 years. His laboratory background is metabolic control and his PhD centered on kinetic regulation of enzyme kinetics and computer simulation. He has been involved in sponsored and investigator-initiated clinical trials and has had committee service at the UCSD Clinical Research Center and on the Clinical and Translational Research Center. He runs the UCSD Biochemical Genetics and Metabolomics Laboratory and has been professionally and scientifically focused on cystinosis since he began working with Dr. Jerry Schneider over a decade ago to modernize intracellular cystine testing.

Bruce Barshop, MD, PhD
Professor of Clinical Pediatrics
University of California, San Diego

RESEARCH · HOPE · CURE

TOGETHER,
WE'RE TURNING

POSITIVITY into PROGRESS!

THANK YOU CANADA

CRF and Canadian Families Unite to Fund Research

Canadian cystinosis families are committed CRF partners working with CRF to fund research that will lead to better treatments and a cure. Families across Canada continue to organize and plan events to raise money for research. Working together, our two countries have united in their efforts to raise awareness about cystinosis, to advocate on behalf of all children and adults with cystinosis and to ensure that we will fund the most qualified researchers in the world.

Since 2016, Canadian families have directly funded CRF research with $688,012 in grant payments through the Aqueduct Foundation and Liv-A-Little Foundation. CRF is incredibly grateful for the support of our Canadian neighbors and friends.

Recent grants paid through Aqueduct:

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

Novel Mechanistic and Translational Studies of Neutrophil-Mediated Inflammation in Cystinosis

$37,500 *Funded through Aqueduct Foundation*

Liang Feng, PhD
STANFORD UNIVERSITY
PALO ALTO, CA

Molecular Mechanism of Cystinosis

$37,500 *Funded through Aqueduct Foundation*

Donations made by Canadians are channeled through Canada Helps, managed by the Aqueduct Foundation and administered by Cystinosis Awareness and Research Effort (CARE). Through Canada Helps, CARE has created an efficient and effective fundraising process, ensuring that Canadians receive a tax receipt. Canadians can also donate directly to CRF. If you would like to learn more about how to fundraise in Canada or to make a donation, please contact: Zoe Solsby *(zsolsby@cystinosisresearch.org).*

AREAS OF RESEARCH FOCUS & GRANTS AWARDED SINCE 2002

THE IMPACT OF CRF RESEARCH

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

32 GRANTS

NEW

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

NEW

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
Zhiyog Chen, PhD
UNIVERSITY OF ZÜRICH, ZÜRICH, SWITZERLAND

Liang Feng, PhD
STANFORD UNIVERSITY, PALO ALTO, CALIFORNIA

NEW

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

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

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

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

Alessandro Luciani, PhD
UNIVERSITY OF ZÜRICH, ZÜRICH, SWITZERLAND

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

Norbert Perrimon, PhD
HARVARD MEDICAL SCHOOL, BOSTON, MASSACHUSETTS

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

NEW

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

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

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

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THE IMPACT OF CRF RESEARCH

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 Levtchenko, MD, PhD
UNIVERSITY HOSPITAL, LEUVEN, BELGIUM

Thyroid

1 GRANT

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

Cure Cystinosis International Registry (CCIR)

1 GRANT

Ranjan Dohil, MD
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

NEW

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 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

AREAS OF RESEARCH FOCUS & GRANTS AWARDED SINCE 2002

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

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

Kidney Research

20 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

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

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

Skin, Muscle and Bone

12 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

NEW

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

Neurological

15 GRANTS

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

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

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

Florian Eichler, MD
MASSACHUSETTS GENERAL HOSPITAL,
BOSTON, MASSACHUSETTS

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

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

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

Genetic Analysis of Cystinosis

5 GRANTS

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

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

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

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

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

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

SPRING 2019 CRF RESEARCH
GRANTS FUNDED

CYSTINOSIS RESEARCH FOUNDATION

Approved for Funding: Six Grants Including Gift to Dr. Cherqui

$1,761,223

1 Ketogenic Diet for Nephropathic Cystinosis

TWO-YEAR STUDY
$200,310 FUNDED

Francesco Bellomo, PhD (PI)
Francesco Emma, MD (Co-PI)
Bambino Gesù Children's Hospital, IRCCS, Rome, Italy

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

TWO-YEAR STUDY
$150,000 FUNDED

Sergio Catz, PhD (Mentor)
Nadia Zgajnar, PhD (Fellow)
The Scripps Research Institute, La Jolla, California

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

TWO-YEAR STUDY
$362,746 FUNDED

Stéphanie Cherqui, PhD (PI)
University of California, San Diego, La Jolla, California

4 Cathepsin D Inhibition to Rescue Osteoblast Function in Cystinosis

TWO-YEAR STUDY
$125,950 TOTAL GRANT AMOUNT
$62,975 FUNDED (ONE YEAR ONLY)

Andrea Del Fattore, PhD (Mentor)
Giulia Battafarano, PhD (Fellow)
Bambino Gesù Children's Hospital, IRCCS, Rome, Italy

5 A Novel Absorptive Epithelium Defect in Cystinosis

ONE-YEAR STUDY
$75,000 FUNDED

Bruno Gasnier, PhD (Mentor)
Yann Teriz, MSc (Fellow)
Paris Descartes University, Paris, France

6 Elucidation of Tissue-Specific Roles of Cystinosin in mTORC1 Signaling and Mitochondrial Metabolism

TWO-YEAR STUDY
$145,000 FUNDED

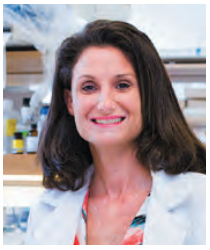
Matias Simons, MD (Mentor)
Marelja Zvonimir, PhD (Fellow)
Institut Imagine, Paris, France

GIFT AWARDED

7 Stem Cell and Gene Therapy Clinical Trial

FOUR-YEAR GIFT
$765,192 FUNDED

Stéphanie Cherqui, PhD
University of California, San Diego, La Jolla, California



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LAY ABSTRACTS
STARTING ON
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Ketogenic diet for nephropathic cystinosis

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

OBJECTIVE/RATIONALE:

Ketogenic diet is a low-carbohydrate/high-fat diet that shifts the body's metabolism away from carbohydrates and towards fat and ketones. Positive effects have been recently described in several conditions, in particular in metabolic and neurological disorders. Ketogenic diet reduces inflammation and improves energy production in cells, which are known to be compromised in cystinosis. The goal of this project is to test the effects of ketogenic diet in cystinotic animals.

PROJECT DESCRIPTION:

In preliminary experiments, we observed that cystinotic mice fed with a ketogenic diet from the age of 3 months to the age of 12 months no longer develop significant kidney disease. We now propose to test if positive effects are also observed if treatment is started later, when renal disease has developed, mimicking the situation in the human disease. In addition, we will analyze the effects of this diet on muscles, since myopathy is a severe late complication of cystinosis. In parallel experiments, we will test if the same results can be obtained with a compound that mimics ketogenic diet, without many of its side effects and will also repeat the above experiments in a new rat model of cystinosis.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

If successful, this project will provide a proof-of-concept allowing testing this approach in patients with nephropathic cystinosis. In particular, treatment with drugs mimicking ketogenic diet could represent a very valuable therapeutic approach in this disease.

ANTICIPATED OUTCOME:

We anticipate confirmation of our preliminary data on renal function and hope that these positive results will be extended to other organs, in particular to the muscular system.

Role of cystinosin in the cross-talk between late endosomal, endoplasmic reticulum and autophagic functions

Sergio D Catz, PhD, *Principal Investigator*
Nadia Zgajnar, PhD, *Research Fellow*
THE SCRIPPS RESEARCH INSTITUTE, LA JOLLA, CALIFORNIA

OBJECTIVE/RATIONALE:

Autophagy, or self-eating, is a process by which the body removes damaged cells and recycles their parts. Preliminary data from our laboratory found that repairing vesicular trafficking improves some of the defective phenotypes related to autophagy in cystinosis. Our working hypothesis is that the lysosomal storage disorder is associated with a decrease or a defective function of multiple trafficking regulators which dysregulate autophagy in cystinosis. The objective of the work proposed is to characterize novel mechanisms of vesicular trafficking associated with autophagy dysregulation in cystinosis.

PROJECT DESCRIPTION:

To understand the molecular mechanisms that are involved in the communication between late endosomal defects and macroautophagy, we will systematically study the effect of trafficking mediators (named Rab GTPases and effectors) and medically relevant cystinosin point mutants (CTNSK208R, juvenile cystinosis; CTNSG308R, infantile cystinosis), and wild-type CTNS, as control, on the cellular localization of molecules that regulate transcription (the reading of the code in the DNA to generate new proteins) in cystinotic cells. We will study the role of a novel molecule, Atg9b, its expression and localization, trafficking and the regulation of autophagic activity. These studies will be carried out using two different types of cystinotic cells: Ctns-/- fibroblast and CTNS-KO HK2 proximal tubule cells (PTCs). The impact of cystinosin function on autophagic mechanisms in inflammatory cells will be studied by validating key results in Ctns-/- neutrophils and macrophages (immune, pro-inflammatory cells).

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

It is expected that the completion of this work will help us understand the newly identified mechanisms regulating the cross-talk between the processes of late endosomal trafficking and macroautophagy. We also expect to clarify the role of cystinosin as a trafficking regulator, and to identify cell-specific phenotypes that will lead to the discovery of new targets for the treatment of cystinosis.

ANTICIPATED OUTCOME:

It is still unclear which are the molecular defects that affect cystinotic cells. We expect to learn how these cells work, identify differences between healthy and cystinotic cells, and identify new molecular targets that can be used to design novel approaches to treat cystinosis.

Mechanism of bone marrow stem cell-mediated therapy in the mouse model of cystinosis

Stéphanie Cherqui, PhD, *Principal Investigator*

UNIVERSITY OF CALIFORNIA, SAN DIEGO, SAN DIEGO, CALIFORNIA

OBJECTIVE/RATIONALE:

The objective of this project is to investigate the mechanism by which transplantation of hematopoietic stem cells (HSCs), which are blood stem cells, can lead to long-term tissue preservation in cystinosis.

PROJECT DESCRIPTION:

Hematopoietic stem cell (HSC) transplantation leads to long-term tissue preservation in the mouse model of cystinosis, the Ctns-/- mice. We showed that HSCs migrate to tissues and differentiate into macrophages that generate long tubular protrusions called tunneling nanotubes (TNTs) that mediate the transfer of "healthy" lysosomes to the adjacent disease cells.

We studied in vitro and in vivo the macrophage phenotype and conditions that inhibit or enhance macrophage-generating TNTs and intercellular organelle trafficking. We reported that macrophage co-culture with diseased Ctns-/- fibroblasts yields increased frequency of membrane protrusions as well as lysosomal and mitochondrial intercellular trafficking, as compared to macrophage co-culture with wildtype (WT) fibroblasts. This demonstrates that secreted components from the Ctns-/- cells enhance TNT formation.

Thus, we will investigate the agents secreted by the diseased cells and involved in TNT formation. In addition, to determine the exact phenotype of the macrophages capable of generating TNTs and responsible for long-term tissue rescue in Ctns-/- mice, we will study the expression profile and regulation of the key genes expressed in the macrophages capable of generating TNTs and involved in the tissue preservation.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

This work will allow the understanding of the mechanism by which HSC transplantation rescues cystinosis. This knowledge will be important for the stem cell gene therapy clinical trial for cystinosis.

ANTICIPATED OUTCOME:

This work also already opened new perspectives in regenerative medicine that spurred the development of novel stem cell-based therapy for other disorders.

Cathepsin D inhibition to rescue osteoblast function in cystinosis

Andrea Del Fattore, PhD, *Principal Investigator*
Giulia Battafarano, PhD, *Research Fellow*
BAMBINO GESÙ CHILDREN'S HOSPITAL, ROME, ITALY

OBJECTIVE/RATIONALE:

Patients affected by cystinosis develop hypophosphatemic rickets. We demonstrated that skeletal alterations and decreased mineralization in cystinotic mice are also due to primary defects of bone forming cells osteoblasts. We demonstrated that cystinotic osteoblasts showed increased expression of the protease cathepsin D. Since it was reported that the administration of cathepsin inhibitors improves bone mineralization in hypophosphatemic cells, this project aims to understand the effects of the cathepsin D alterations in cystinotic osteoblasts and to evaluate whether its inhibition could restore the physiological bone remodeling.

PROJECT DESCRIPTION:

This project aims to evaluate the role of cathepsin D in the reduced osteoblast differentiation and activity observed in cystinosis. We will analyze in vitro the effects of cathepsin D inhibition on cystinotic osteoblasts with the goal to restore the physiological bone formation. To test our hypothesis we will treat osteoblasts with cathespin D inhibitor and we will evaluate the effects on their differentiation and activity. Since osteoblasts regulate the differentiation and activity of bone resorbing cells osteoclasts to maintain the integrity of the skeleton, we will also analyze the osteoclastogenic potential of treated cystinotic osteoblasts. These experiments will be important to test whether cathepsin D inhibition could re-establish the correct bone remodeling.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

The project proposes to characterize the role of cathepsin D in the cystinotic skeletal disease and to understand whether cathepsin D inhibition could restore the osteoblast differentiation and mineralization impaired in cystinotic cells. This study will be important to identify a potential target for the treatment of skeletal alterations occurring in cystinosis with the long-term goal to translate the results into benefits for patients.

ANTICIPATED OUTCOME:

The *in vitro* study will allow us to better understand the role of cathepsin D in cystinotic bone pathology. By blocking the cathepsin D activity into cystinotic osteoblasts, we expect to discover if its inhibition can restore the normal functions such as differentiation, mineralization ability and cross-talk with osteoclasts. These results will open the way to test whether cathepsin D-targeted treatment could restore the physiological bone remodeling activity in an animal model of cystinosis.

A novel absorptive epithelium defect in cystinosis

Bruno Gasnier, PhD, *Principal Investigator*
Yann Terres, MSc, *Research Fellow*
PARIS DESCARTES UNIVERSITY, PARIS, FRANCE

OBJECTIVE/RATIONALE:

Our genetic studies of the mouse model of cystinosis showed that cystinosin functionally interacts with another lysosomal protein during early embryonic development. We found that this interaction occurs in the yolk sac, an extra-embryonic organ which shares functional similarities with the kidney proximal tubule, a structure severely affected during infancy in cystinosis. Loss of the two proteins leads to drastic defects in yolk sac intracellular membranes, suggesting a novel cellular function of cystinosin. Our goal is to unveil this novel function and to assess its potential significance for the pathogenesis of cystinosis.

PROJECT DESCRIPTION:

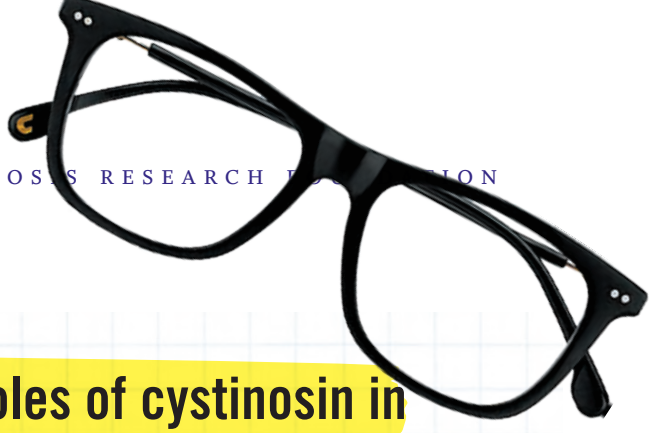
We will further analyze the morphological defects of cystinosin-deficient yolk sac cells in distinct genetic backgrounds and investigate the molecular and cellular basis of these defects using complementary analytical techniques. Diverse cellular pathways will be analyzed in normal and mutant yolk sacs using microscopy and biochemical approaches. We will also assess the ability of yolk sac cells to internalize and degrade proteins, a function shared with the kidney proximal tubule, using dedicated fluorescent probes. Treatment of acutely isolated yolk sacs with cysteamine or with other drugs will tell whether the intracellular defects are caused by defective lysosomal cystine export or by the loss of other functional features of cystinosin. Finally, we will investigate the causal relationship between the yolk sac defects and the embryonic defects using genetic rescue experiments.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

The yolk sac cells studied in this project and kidney proximal tubules share several functional and biochemical features, including the active internalization and degradation of extracellular proteins. Kidney proximal tubules are rapidly and severely affected in cystinosis. However, in our mouse models, the cellular defects develop much faster in yolk sac cells than in kidney proximal tubules (days versus months). Therefore, the yolk sac pathology may represent a “superfast” model to investigate the pathogenesis of cystinosis.

ANTICIPATED OUTCOME:

This project should unveil novel aspects of the cell biology of cystinosin, and potentially novel mechanisms at work in cystinosis. This novel function surprisingly requires cooperation with another lysosomal protein, suggesting novel potential targets for therapeutic intervention. As the yolk sac epithelium shares functional and mechanistic similarities with kidney proximal tubules, our study may help understand the Fanconi syndrome of cystinosis and its apparent insensitivity to cysteamine therapy.



Elucidation of tissue-specific roles of cystinosin in mTORC1 signaling and mitochondrial metabolism

Matias Simons, MD, *Principal Investigator*
Marelja Zvonimir, PhD, *Research Fellow*
IMAGINE INSTITUTE, PARIS, FRANCE

OBJECTIVE/RATIONALE:

By using fruit flies (Drosophila) as a model, we discovered a novel role for cystinosin (CTNS) in a metabolic pathway, in which the amino acid cysteine limits too high mTOR reactivation in extended periods of starvation. This pathway is essential for maintaining autophagy, which describes the self-eating of cells when external nutrients are lacking. We discovered high activity of this pathway in fat bodies, a tissue analogous to liver/adipose tissue in mammals. However, intriguing differences with regard to mTOR signaling, autophagy and mitochondrial metabolism were observed in Drosophila nephrocytes, cells that fulfill functions analogous to mammalian podocytes and proximal tubular cells. We therefore believe that CTNS also has a tissue-specific function with regard to the mTOR pathway.

PROJECT DESCRIPTION:

We will use both established and newly generated fly tools to unravel the tissue-specific roles of CTNS in *Drosophila* fat bodies and nephrocytes with regard to mTOR signaling, autophagy and mitochondrial function. Secondly, we aim to validate these roles in mammalian cell model systems. We humanized flies by replacing the fly CTNS with human CTNS variants carrying different point mutations identified in cystinosis patients. Our preliminary data argue for an additional scaffolding function of CTNS in nephrocytes to its already existing metabolic role as supplier of cytosolic cysteine. We will perform rescue experiments to prove whether the metabolic function of CTNS, discovered in fat body cells, is also present in nephrocytes, masked by the additional nephrocyte-specific scaffolding function. These studies will allow a deeper understanding of the tissue-dependent roles of cystinosin.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

Experiments will prove whether the metabolic function of CTNS discovered in the fly liver also exists in nephrocytes and whether it is masked by an additional nephrocyte-specific CTNS scaffolding function. Thus, our newly discovered CTNS-dependent cysteine catabolism pathway might be crucial for certain cell types to cope with fasting periods or other nutrient-dependent fluctuations in mTOR signaling whereas other cells have additional layers of regulation to prevent that these fluctuations affect mTOR signaling. Unravelling these tissue-specific roles of CTNS might be important for targeted treatment strategies.

ANTICIPATED OUTCOME:

In flies, fat body cells and nephrocytes both express CTNS but intriguing differences with regard to mTOR signaling, autophagy and mitochondrial dynamics are shown when CTNS is mutated. Finding proof that CTNS has tissue-specific roles will be useful to develop more specific treatments for cystinosis.

CALL FOR FALL 2019 RESEARCH PROPOSALS

RESEARCH IS OUR HOPE

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 16 short years. Since its inception, CRF has funded 187 multi-year research studies in 12 countries. Our researchers have published 80 articles in prestigious journals as a result of CRF funding. Every dollar donated goes directly to support cystinosis research.

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

In September, CRF announced $2.5 million was available for the Fall 2019 call for research proposals and fellowship grants. The grant awards will be announced at the end of December 2019.

In Spring 2019, CRF issued 7 new grants for over $1.7 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.

CCIR

In 2010, CRF established the **Cure Cystinosis International Registry (CCIR)** to serve as a hub of information about cystinosis and its complications. Currently, CCIR has 576 registrants from 44 countries. The site, which includes a Professional Research Portal, is a critical resource for researchers and scientists who register to access and view de-identified, aggregate cystinosis patient information. The portal can be accessed at *www.cystinosisregistry.org*.

www.cystinosisresearch.org/research/for-researchers

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.

SAVE THE DATE

SEVENTH INTERNATIONAL

CYSTINOSIS RESEARCH SYMPOSIUM

Sponsored by the Cystinosis Research Foundation

THURSDAY, FEBRUARY 27, 2020
– AND –
FRIDAY, FEBRUARY 28, 2020

Arnold and Mabel Beckman Center of the National Academies of Sciences and Engineering
IRVINE, CALIFORNIA

WE ARE HONORED TO ANNOUNCE
OUR KEYNOTE SPEAKER

Morton J. Cowan, MD

University of California, San Francisco
Allergy, Immunology, and Blood and Marrow Transplant Division

— PRESENTING —

CD34+ Hematopoietic Stem Cell Gene Therapy For Severe Combined Immunodeficiency Disease

2020 SYMPOSIUM CO-CHAIRS

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Stéphanie Cherqui, PhD

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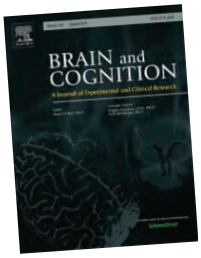
BY INVITATION ONLY

cystinosis RESEARCH FOUNDATION | TOGETHER WE SHINE BRIGHT

For Information contact CRF President Nancy Stack
949.223.7610 or nstack@cystinosisresearch.org

PUBLISHED STUDIES

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



Since 2004 CRF has provided grant funding to ***Doris A. Trauner, MD, University of California, San Diego****, for her important work on cystinosis and the brain. Dr. Trauner was recently published in the* Brain and Cognition Journal *for her research on* **VISUAL AND VERBAL LEARNING AND MEMORY IN CYSTINOSIS**.

CENTRAL NERVOUS SYSTEM COMPLICATIONS IN ADULT CYSTINOSIS PATIENTS, *published August 2019 in the* Journal of Inherited Metabolic Disease *by* ***Aude Servais, MD, PhD, IMAGINE Institute of Genetic Diseases, Necker Hospital, Paris, France.***

MACROPHAGE POLARIZATION IMPACTS TUNNELING NANOTUBE FORMATION AND INTERCELLULAR ORGANELLE TRAFFICKING, *published October 2019 in* Scientific Reports Nature Research Journal *by* ***Stéphanie Cherqui, PhD, and Spencer Goodman, Graduate Student Researcher, University of California, San Diego.***

LRP2 REGULATES PROINFLAMMATORY AND ANTIAPOPTOTIC RESPONSES IN PROXIMAL TUBULAR EPITHELIAL CELLS, *published October 2019 in* Frontiers In Cell and Development Biology *by* ***Giusi Prencipe, PhD, Bambino Gesù Children's Hospital, Rome, Italy.***

TOGETHER

We're Turning Positivity Into *Progress*

natalie's

wish20

Saturday, April 18, 2020

6:00 pm Cocktail Reception
7:15 pm Program and Dinner

Honoring the children and adults who are affected by cystinosis and the cystinosis research community for its commitment to our children.

where

Fashion Island Hotel
690 Newport Center Dr,
Newport Beach, CA 92660

featuring

Matt Mauser
his Sinatra Big Band and
The Tijuana Dogs

cystinosis RESEARCH FOUNDATION

cystinosisresearch.org

together

we're turning hoping into happening

day of hope20

Learn, share, laugh and celebrate for three inspiring days with fellow members of the cystinosis community. CRF-funded researchers and clinicians will lead discussions on stem cell and gene therapy, treatments for corneal cystinosis, muscle wasting and myopathy, kidney disease and more.

The conference will include important sessions led by CRF-funded researchers and clinicians.

Confirmed speakers include:

Stéphanie Cherqui, PhD
University of California, San Diego
San Diego, CA

Morgan Fedorchak, PhD
University of Pittsburgh School of Medicine *Pittsburgh, PA*

Benjamin "Beno" Freedman, PhD
University of Washington,
Seattle, WA

Paul Grimm, MD
Stanford University School of Medicine
Palo Alto, CA

Larry Greenbaum, MD, PhD, FAAP
Emory School of Medicine
Atlanta, GA

Stephen Jenkins, MD
University of Utah School of Medicine
Salt Lake City, UT

when

Thursday, April 16, 2020 – Saturday, April 18, 2020

where

Fashion Island Hotel
690 Newport Center Dr,
Newport Beach, CA 92660

For more information on the conference or the Family Assistance Fund contact:
info@cystinosisresearch.org
949.223.7610

cystinosis RESEARCH FOUNDATION

cystinosisresearch.org

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!

Olivia Little – Port Elgin, Ontario, Canada

SWING, SHOOT AND LIV GOLF CLASSIC

On Saturday, September 14 the sun was shining at the Saugeen Golf Club in Saugeen Shores when 216 golfers gathered to hit the greens to support the 7th Annual Swing, Shoot and Liv Golf Classic held in honor of Olivia Little. Another stellar event that wouldn't have been possible without the 30 volunteers that made the day happen.

Olivia and her sister Harper got the golfers started with their signature tee-offs. Teams quickly got in their carts and scattered across the 27-hole course where they would find different venues and fundraising events throughout the course. The most talked-about was the putting challenge, where golfers were challenged to make a 30-foot putt into a clawfoot tub. The clawfoot tub with random holes drilled into it was meant to educate golfers about what cystinosis does to Olivia's kidneys.

Following the tournament, golfers shared a delicious meal and competed against each other in the silent and live auction. Katelynn Campbell from Wallaceburg and I spoke and opened our hearts as mothers of children with cystinosis, giving the room a glimpse of "the good, the bad and everything in between." At the end of the night, we raised $120,000 for cystinosis research. We are so proud of our community and the love they have for Liv. We truly are better together.

TOGETHER, WE ARE one

Kenzie Lawatsch — Coleman, Wisconsin

3RD ANNUAL DRIVING FORE A CURE

The Lawatsch family held their 3rd Annual Kenzie's Driving Fore a Cure golf tournament on a beautiful day in August. The amazing weather set the tone for a fun-filled afternoon of golf, surrounded by our amazing family and generous community of friends.

Words just aren't enough to express our appreciation for our sponsors who have always been so generous and supportive, and for all the golfers who make the day so special with their golf stories and comradery. And a special thank you to those who donated items for the raffle and prizes and especially to our volunteers who make the day a success.

Each of you has made a difference by raising $8,114 for cystinosis research in support of Miss Kenzie in the fight for a cure, thank you!

Aidan O'Leary – Chicago, Illinois

AIDAN'S ARMY GOLFS FORE A CURE

Another win for Aidan's Army! Over 100 friends and family members from across the country joined the fight to cure cystinosis on July 29, 2019, at Forest Lake Country Club in Bloomfield Hills, Michigan. Aidan's Army really showed strength in numbers after they raised over $73,785 for cystinosis research. Erin and Jim O'Leary are forever grateful to their amazing donors and cannot wait to host the event again next year! Thank you so much, O'Leary family and Aidan's Army, for your generous contributions. They will be used to fund recently awarded research grants focused on improving the lives of those with cystinosis!

AIDAN'S ARMY

1 PURPOSE. 1 JOURNEY. 1 CURE.

Hayden Kirchhof – Denver, Colorado

WINE & CHEESE RECEPTION IN HONOR OF HAYDEN

In late spring, Angela and Nick Kirchhof hosted their first Wine and Cheese Reception in honor of their daughter Hayden and it turned out to be an enormous success. Over 160 enthusiastic guests attended the event bidding on 49 auction items. It was an incredible night of celebration and community. Together, their family and friends raised $36,200 that night for cystinosis research and to honor Hayden.

Since Hayden was diagnosed in November 2018, CRF has received a total of $79,860 in Hayden's honor. On behalf of CRF and the cystinosis community, thank you for your hard work and commitment to help fund cystinosis research for a better future for our children. Together we are changing the face of cystinosis forever, thank you!

Andrew Cunningham – Langdon, Alberta, Canada

FOREFATHERS GOLF TOURNAMENT

The 8th Annual JCFG Memorial Golf Tournament was held at the Boulder Creek Golf Course in Langdon, Alberta, Canada in honor of Andrew Cunningham who was diagnosed with cystinosis at 17 months of age. Andrew, now 15 years old, is in grade 11 and a happy teenager.

With the support of our sponsors, amazing volunteers and enthusiastic community the tournament was another memorable day of golf with Andrew's uncles, Alan and Neil, donning outrageous costumes as they made their way around the course entertaining the golfers with their usual shenanigans.

The event started in 2012 as a tribute to four fathers, John McCullagh, Conway Cameron, Frank Halluk & Gordon Cunningham (JCFG) who lost their lives to heart disease. John and Gordon were Andrew's granddads, so in addition to raising funds for heart disease, proceeds from the event also support cystinosis research. We are so proud of the partnership we have with the CRF and will continue to drive the funds we raise, in support of the incredible efforts of the doctors and researchers dedicated to battling this disease alongside our families. We have full faith that one day, the tournament will be held in celebration of the cure of cystinosis!

McCullagh and Cunningham Family

TOGETHER, WE ARE one

Charlotte Coe — Webster, New Hampshire

MAST YARD TRAIL RACE HONORS CHARLOTTE COE OF NEW HAMPSHIRE

Each year the Mast Yard Trail Race held in Contoocook, New Hampshire, raises money for a compelling charity to receive 100% of the proceeds from the race. This year in honor and support of a local New Hampshire child, Charlotte Coe, we selected the Cystinosis Research Foundation as the charity.

The event, held in July, brought out 65 enthusiastic runners to support our cause. We would like to thank our #1 sponsor Northeast Delta Dental. Thank you also to Stoneyfield Yogurt, Cracker Barrel and Best Septic for their generosity and support!

The race was a huge success and brought awareness to the participants of the tremendous challenges of cystinosis that face Charlotte and her family. We were honored to have raised approximately $1,750 which was sent to CRF for recent cystinosis research grant funding. Thank you to all who came out for race day to support the Coe Family and the charity.

Ellen Raffio, Race Director

Abbi Monaghan — St. Catharines, Ontario, Canada

TARGET CYSTINOSIS – DECEW GUN CLUB HOSTS SHOOT FOR ABBI MONAGHAN

Katie and Terry Monaghan of St. Catharines, Ontario, Canada, were overwhelmed by their community's support of the 4th Annual Shoot for Abbi fundraiser held in honor of their daughter Abbi. Each year the event has been hosted by the Decew Gun Club and organized by John Rakich and his family. The volunteers and attendees helped to raise more than $15,660 to support important cystinosis research. On behalf of CRF and the cystinosis community, thank you to the Monaghan and Rakich families and the Canadian community for helping to ensure a brighter future for those affected by cystinosis. Together we shine bright!

1 PURPOSE. 1 JOURNEY. 1 CURE.

Landon Hartz — Pittsburgh, Pennsylvania

HARTZ FAMILY CHARITY GOLF OUTING FOR LANDON

On behalf of Landon and the rest of our family, we wanted to thank you for participating in the 8th Annual Lots of Love for Landon Charity Golf Outing. Having a child that is born with a disease is difficult for any family to endure. Organizing charitable events allows the parents and families of the child the opportunity to feel like they are doing as much as they can to contribute to the child's quality of life. The kindness and generosity of people like you are what really makes the events successful though!

We're extremely excited to announce that we were able to raise over $25,000 this year! This brings our overall total to over $150,000!!! Thank you to those of you that have been with us since the first year, and to the newcomers for allowing us to continue to grow every year. Every dollar that was raised goes directly to the Cystinosis Research Foundation to help fund research that will result in better medical care and we hope ultimately a cure.

Hopefully, you enjoyed some of the new features this year (the Early Bird Special, the Blue Ball Game, etc.). We'll look to continue these and see what else we can try next year to keep it fun and entertaining for everyone. We are going to make a strong push to get just 10 more groups than we had this year so we can fill the entire 36-hole course, which would be a great accomplishment considering where we started 8 years ago. Please invite your friends to join in the fun in 2020!

Morgan Peachman — Avon Lake, Ohio

MORGAN PEACHMAN'S EXTENDED TMC FAMILY CONTINUES SUPPORT OF CRF

When I joined The Mortgage Collaborative nearly 4 years ago, like any mother in a new job with a medically fragile child, I was hesitant at first to share Morgan's diagnosis. However, when you're running your family's charity golf outings word tends to leak out. After our President and Vice Chairman had the opportunity to meet Morgan in Cleveland for one of our Mulligans Fore Morgan events, he decided right then and there to make our company's bi-annual golf outing charity events to support the Cystinosis Research Foundation.

This year has been a challenging one for our family, as we're now preparing for Morgan's first kidney transplant. After three surgeries this year already, I ended up having to leave TMC's Winter Conference early to fly home due to complications. While I was gone, David raffled off a bottle of Pappy Van Winkle to boost funds for Morgan's medical fund during our time of need. As we prepare for me to donate my spare kidney to my girl, we unfortunately, made the decision to cancel our family's golf outing so we can focus on her care as her health continues to decline.

The Mortgage Collaborative has stepped up in our stead with their support, hosting two Mulligans Fore Morgan golf outings in Austin and Nashville. This year, we've raised over $15,000 from our TMC Family for the Cystinosis Research Foundation. And plans are already underway for two more events in New Orleans and Denver in 2020!

TOGETHER, WE ARE one

Sophie Betournay – Kanata, Ontario, Canada

HOUSE OF LUCK FOR CYSTINOSIS

On May 11, 2019, the Betournay-Chatelain family from Ottawa, Ontario, Canada hosted their first-ever fundraising event in honor of Sophie Betournay. The "House of Luck for Cystinosis" casino night event was a smashing success, pulling in $2,500 in donations for CRF! The event was held at the Art House Café, the downtown Ottawa art gallery and trendy café that Sophie's aunt Genevieve runs with the help of her wonderful staff. Many local businesses and generous friends and family donated amazing prizes to this event. The family thanks everyone who helped make it the incredible night that it was - planning for 2020 is already underway!

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! As of October 7, our Facebook Friends have raised over $43,500 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 2019 fall grant awards are announced! To get started, go to this web address: *www.facebook.com/fund/CystinosisResearchFoundation*.

Jake Krahe — Medina, Ohio

PETROS HOMES GOLF TOURNAMENT IN HONOR OF JAKE KRAHE

As a homebuilder, all our business is done on a local level. The homes we build and the people who build them are all part of the local community.

Throughout the years we hear many stories of need or causes worth supporting that are connected to our employees or trade and supply partners. Twelve years ago, we started a golf outing to help benefit or support causes with a connection to our community. This year we were honored to hold the golf outing in support of the Cystinosis Research Foundation. The foundation was chosen because of the Krahe Family in Medina, Ohio, whose son Jake has cystinosis.

The support we receive from our sponsors who are made up of our trade, supply, banking and manufacturing partners is always humbling. Everyone is so eager to help someone within our community who is in need and it is always a blessing to see who the funds are going to support. Our prayer is that these funds along with the generosity of others will be combined to provide healing and hope to those who are suffering from this terrible disease. Petros Homes is honored to contribute $10,000 in Jake's name to support the Cystinosis Research Foundation and their grant funding to find better treatments and a cure for cystinosis.

Carla Santora – Broadview Heights, Ohio

1 PURPOSE. 1 JOURNEY. 1 CURE.

Barbara Tschannen – Brookfield, Missouri

WESTON TSCHANNEN MEMORIAL TOURNAMENT

We hosted the third annual Weston Tschannen Memorial Golf Tournament on Saturday, May 18, 2019, at the Brookfield Country Club, Brookfield, Missouri. We had 42-hole sponsors, 130 golfers, and lots of items for the live auction that is held at the end of the tournament. This is a fun day of golf supported by our community, along with Weston's family and friends.

We have been very blessed to have a wonderful turnout every year. There were storms predicted on that Saturday, but the rain held off just long enough to finish the 18 holes and allow everyone to get into the Pro Shop for the auction. We would like to think Weston had a hand in that good fortune!

Overall, the tournament was a huge success, raising over $22,000 for cystinosis research and donations to local organizations that Weston was passionate about, such as wrestling, the YMCA and the animal shelter. The support we receive in Weston's memory is so comforting to our family!

Jenna and Patrick Partington – Sacramento, California

CAPITAL CUP GOLF TOURNAMENT

Jenna and Patrick are pictured here on September 29, 2019 with Sacramento Capital Cup tourney participants Shannon Deary Bell of NorCal Beverage (raised $28,000) and Tom Walcott of Colliers International (raised $12,000). We thank these two community leaders and dear friends for raising funds and playing on behalf of Jenna & Patrick's Foundation of Hope! We love you, Shannon and Uncle Tommy!

Sam and Lars Jenkins – Salt Lake City, Utah

HAUNTED HOUSE HOSTED BY SAM AND LARS

Sam and Lars Jenkins have been dying to have a haunted house, and on Saturday, October 12th, they hosted a spooky haunted Halloween house in their home. They charged a $1 entry fee and sold hot chocolate and donuts. All money raised went to support the Cystinosis Research Foundation. At the end of the evening nearly $1,000 was raised for cystinosis research. Thank you Sam and Lars!

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, November 25, 2019

KEGS 4 KAUSE, IN HONOR OF HADLEY ALEXANDER
Payette Brewing Co., Boise, Idaho
For information contact Marcu Alexander, hearts4hadley@gmail.com

Wednesday, December 4, 2019

JOLLY FOR JOSIE IN HONOR OF JOSIE KANUPKE
Hailstorm Brewery, Tinley Park, Illinois
For information contact Laura, lmchristophers30@gmail.com

Saturday, December 28, 2019

GRAND GALA CELEBRATION IN HONOR OF AARAV KHALASI
The Sutter Club, 1220 9th St. Sacramento, California 95814
For information contact Mukund Khalasi, mukund.khalasi@icloud.com

Saturday, March 28, 2020

2400FT OF SCHWEITZER, 24 HOURS FOR HANK, HENRY STURGIS
Schweitzer Mountain, Sandpoint, Idaho
For information contact information@24hoursforhank.org

Friday, May 29, 2020

LOTS OF LOVE FOR LANDON CHARITY GOLF OUTING, IN HONOR OF LANDON HARTZ
Black Hawk Golf Course, Beaver Falls, Pennsylvania
For information contact LotsofLove4LandonCRF@gmail.com

Summer 2020

AIDAN'S ARMY GOLFS FORE A CURE TOURNAMENT, IN HONOR OF AIDAN O'LEARY
Bloomfield Hills, Michigan
For information contact Katie Emerine, (248) 225-8209 or kcotantemerine@gmail.com

Summer 2020

PAINT THE TOWN PURPLE, IN HONOR OF OLIVIA LITTLE
Port Elgin, Ontario, Canada
For information contact Erin Little, 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, Erin.Little@livalittlefoundation.com

LEADERSHIP. GUIDANCE. COMMITMENT.

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cystinosis RESEARCH FOUNDATION

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 $54 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.

★★★★ CHARITY NAVIGATOR Four Star Charity

Together, *we are*

hope *ing*
go *ing*
lead *ing*
dream *ing*
achieve *ing*
seek *ing*
act *ing*
impact *ing*
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