cystinosis magazine

FOR FRIENDS *and* SUPPORTERS *of the* CYSTINOSIS RESEARCH FOUNDATION

WINTER 2025

WE'RE BETTER TOGETHER

2003 — 2025

Making the journey with HOPE.

One breakthrough after another.

2003 — 2025

Making our journey together with HOPE!

See highlights of our 22-year journey on page 10.

WE'RE BETTER TOGETHER

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 $73 million for cystinosis research in an effort to find a cure.

cystinosis RESEARCH FOUNDATION

Cystinosis Research Foundation
19200 Von Karman Ave. Suite 920
Irvine, California 92612
949 223 7610

CYSTINOSISRESEARCH.ORG

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

To view this edition digitally, visit *www.cystinosisresearch.org/cystinosis-magazine*

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

Project Management: Idea Hall
Art Direction and Design: Born Primate Co.
Printing and Distribution: Goodwork

CYSTINOSIS MAGAZINE IS A PUBLICATION OF THE CYSTINOSIS RESEARCH FOUNDATION

The entire cost to produce and distribute *Cystinosis Magazine* is underwritten by an anonymous donor.

WINTER 2025

IN THIS ISSUE

WELCOME

ANNOUNCEMENTS

FAMILY PARTNERS

RESEARCH UPDATES

Making our journey together with HOPE.

DECEMBER 2025 A LETTER FROM NANCY AND JEFF STACK

Dear Family & Friends,

This is the time of the year when we reflect on our accomplishments and express our gratitude to all of you who are part of the CRF family. Since our humble beginnings in 2003, we have come so far together. As a result of our research program, we have changed the course of cystinosis. Cystinosis, once an unheard-of disease with no steady or significant funding source, now has a continuous source of research funding because of your unwavering support. We have created a synergistic global research community of hundreds of dedicated researchers working every day to find better treatments and a cure for cystinosis.

Natalie's wish "to have my disease go away forever" was the beginning of the CRF story. From that simple and innocent wish grew a remarkable and productive foundation that has become the hub of international research and family connections. The road to a cure has been a long journey, but it has always been guided by hope. Every study we fund and every clinical trial we support brings us closer to the cure. It is your unwavering commitment to our community that has made this progress possible.

UPDATE ON THE NOVARTIS CYSTEM CLINICAL TRIAL

We are closer than ever to realizing the cure for cystinosis. As you know by now, Novartis has opened the next phase of the CYStem cell and gene therapy clinical trial. The CYStem trial is open to children ages 2 – 5 years. The clinical trial will assess the safety, tolerability, and efficacy of the stem cell treatment. Now, some of our youngest patients will be able to participate in this ground-breaking clinical trial. Currently, there are four clinical sites open; two are recruiting for cohort 1 and cohort 0, and two sites are recruiting for cohort 0.

This is another milestone, another giant step forward for our small community. We have worked together, stayed united in our effort to find a cure, and we have driven the research to ensure that this day would be a reality for our community. Here we are on the brink of a new therapeutic treatment that we anticipate will slow the progression of cystinosis or be the cure.

There are two sites open for cohort 0. There is a site at Emory University in Atlanta, Georgia, which is led by Dr. Larry Greenbaum, the Principal Investigator, and a site at Baylor College of Medicine – Texas Children's Hospital, which is led by Dr. Ewa Elenberg. Patients in this cohort (cohort 0) will not receive the stem cell and gene therapy treatment; instead, they will continue their standard of care for cystinosis. Participants in cohort 0 will provide essential context for interpreting the results observed in cohort 1, whose participants will receive the stem cell treatment.

Currently, there are two sites recruiting for cohort 0 and cohort 1. Patients enrolled in cohort 1 will receive the stem cell and gene therapy treatment. The sites include Stanford University, led by Dr. Alice Bertaina and a site at UC San Diego, led by Dr. Nadine Benador. A complete list of the inclusion/exclusion criteria and more information about the sites can be found on the CRF website and on *clinicaltrials.gov.*

If the Novartis trial proves successful, it is anticipated that Novartis will seek accelerated approval for the treatment, making it available for all people living with cystinosis.

The Novartis clinical trial is a reality because of Dr. Stéphanie Cherqui whose passion and commitment for this community continues today. In 2007, CRF awarded Dr. Cherqui its largest grant to date, believing that her vision for a stem cell therapy to treat cystinosis might be a cure for cystinosis. In 2018, Dr. Cherqui received FDA approval to treat six adults with cystinosis and that led to where we are today, on the threshold of treating some of our youngest patients with this groundbreaking therapy.

We are so thankful to the five adult patients who participated in the UC San Diego trial led by Dr. Cherqui. It was because of their participation in the first trial that we have a clinical trial today for younger patients.

CRF LEADERSHIP AND VISION

In partnership with other stakeholders, we have successfully reached this extraordinary milestone. We thank Novartis' stem cell and gene therapy group for their commitment to the cystinosis community. We will continue to work with Novartis to represent the community and to ensure the treatment will be available for all those with cystinosis.

The person we are most grateful to and to whom we owe so much is Dr. Cherqui, who has been our guiding light and a beacon of hope for the entire cystinosis community. We are deeply thankful for her vision, her perseverance, and her extraordinary dedication to the community which continues to drive us forward.

CYSTINOSIS MAGAZINE HIGHLIGHTS

We know you will enjoy this issue of the magazine. We are excited for you to meet Aude Servais, MD, PhD, from Hospital Necker in Paris, France, who is a brilliant researcher and a member of the prestigious CRF Scientific Review Board. You will learn more about the important research she does which focuses on neuromuscular complications in cystinosis. Dr. Servais also shares her vision about where cystinosis research is today and where it is going in the future.

As always, the most joyful and beautiful stories come from our families and patients who live with cystinosis. Each family or patient story has a unique perspective on living with cystinosis and each story reminds us that we are not alone; we are a united community and we have each other to lean on and for support.

>>>>

CRF KNOWS CYSTINOSIS!

We have built a strong, research-driven foundation that has accomplished major milestones, including funding the research that led to two FDA approvals: Procysbi® (2013) and the Stem Cell and Gene Therapy Clinical Trial at UC San Diego (2018). With your support, we have also launched numerous clinical trials and advanced significant discoveries about cystinosis.

Since 2003, CRF has funded 252 multi-year research studies in 13 countries. Our researchers have published 115 articles in prestigious journals because of CRF funding. CRF is the largest private fund provider of cystinosis research in the world. Our policy of awarding research grants twice a year guarantees that there are no gaps in funding and essential and life-saving research is prioritized. Our research program is led by our prestigious CRF Scientific Review Board who carefully and thoroughly evaluate every research application we receive and recommend the most promising research studies to fund.

We are pleased to report that 2025 was another exemplary year for our research program. We funded new researchers who have novel therapeutic ideas and we have continued funding research projects that are on track to yield new discoveries about cystinosis and lead to clinical trials for new treatments. In the spring of 2025, CRF awarded four grants, three new research grants and one lab equipment grant totaling $559,194. The research studies focus on the cellular and molecular aspects of cystinosis, and measuring cystine in the skin using confocal microscopy as a biomarker. We are excited to share the lay abstracts of the new research grants awarded (see page 37).

CRF RESEARCH IMPACTS OTHER DISEASES AND DISORDERS

As we begin the next phase of the stem cell trial, it is important to recognize that the research we fund reaches far beyond cystinosis. The discoveries made by our researchers have advanced research in other diseases including Friedreich's ataxia, Danon disease, Alzheimer's disease, and other genetic and systemic diseases like cystinosis.

Although CRF's mission is to find better treatments and a cure for cystinosis, we are delighted to know that our work has positively impacted other disease communities who are now creating pathways to cures for their communities. Together we are changing lives and giving hope to people far beyond the cystinosis community.

WELCOME TO THE WORLD ISABELLE!

It is with overwhelming joy that we announce the birth of Isabelle Christina Morgan, the beautiful daughter of Natalie and Danny, born on July 24, 2025. Our new chapter as Nana and Grandad to Isabelle is the greatest gift and honor of our lives.

We were thrilled when Natalie and Danny moved back to California from Chicago earlier this year – it has allowed us to be close and to celebrate this new life and new beginning with them.

When I hold Isabelle, I see Natalie's face and I am reminded of how perfect the world can be. Isabelle is full of wonder and innocence, grace and beauty. She has already filled our hearts with joy and love. We are truly blessed and will embrace every moment we have with her.

WITH IMMENSE GRATITUDE

We have so much to be grateful for – your unwavering support, your commitment to research, and your compassion and love for our community. Together we have built a global research community dedicated to improving the lives of children and adults with cystinosis.

Our mission continues and with your support, we will drive research, make new discoveries about cystinosis, launch new clinical trials, and accelerate progress to improve the quality of life for our children and adults with cystinosis. With your continued partnership, we will reach new heights, achieve new breakthroughs, and reach new milestones.

Thank you for being part of the CRF family, and for your support of Natalie and all those with cystinosis. Your commitment to our mission gives us hope that a cure for cystinosis will be a reality soon.

With heartfelt thanks and gratitude,

Nancy and Jeff

2025 has been full of many changes in my life. Earlier this year, I moved back to Southern California with my husband, Danny, and three-year-old dog, Wesley. We wanted to be closer to family as we started a new chapter in our lives.

We bought a beautiful home and love our new neighborhood. It is full of festive decorations for the holidays, and we love going on walks in the neighborhood. Our house has a great backyard for Wesley, who enjoys running around the yard and meeting his furry friends in the neighborhood. In addition to moving back to California, my husband started a new job close to home and I began working again at Court Appointed Special Advocates coaching volunteers on advocating for children in foster care. We have been enjoying being back in sunny California and seeing family and friends more often.

On July 24, 2025, we welcomed our beautiful baby girl, Isabelle Christina Morgan, into the world. She weighed 7 pounds and 2 ounces and was 20 inches long. My husband and I travelled to the Midwest a week before her birth anxiously awaiting her arrival. We hoped she would come a few days early, but she decided to come right on time! We tried to distract ourselves from the wait by exploring the town and meeting our gestational surrogate for meals. Our gestational surrogate did an amazing job taking care of our baby for nine months and we cannot thank her enough for what she did for us. It was a very emotional week and one of the most beautiful moments of my life. We are forever grateful to her and blessed to have our miracle baby with us.

Isabelle is now three months old and getting bigger every day. She loves to wiggle around, coo and smile at us. She already has a little personality and looks like both my husband and me! I could not have asked for a more perfect daughter, and I cannot wait to watch her grow up. I hope that I can be there for her at every big milestone in her life and more than ever now, I want to and need to prioritize my health. With support from the cystinosis community and advanced medical treatments, I have no doubt that I will be strong enough and healthy enough to watch her grow and witness her life milestones.

My medical update is always interesting! I am 34 years old and doing well aside from my kidney function. Last year, my kidney function really started to decline. I see my nephrologist often and get blood work done every three months to monitor the decline of my kidneys. I am currently on the active kidney transplant waitlist. I will likely need a transplant in early 2026, and I am hoping to avoid dialysis before then.

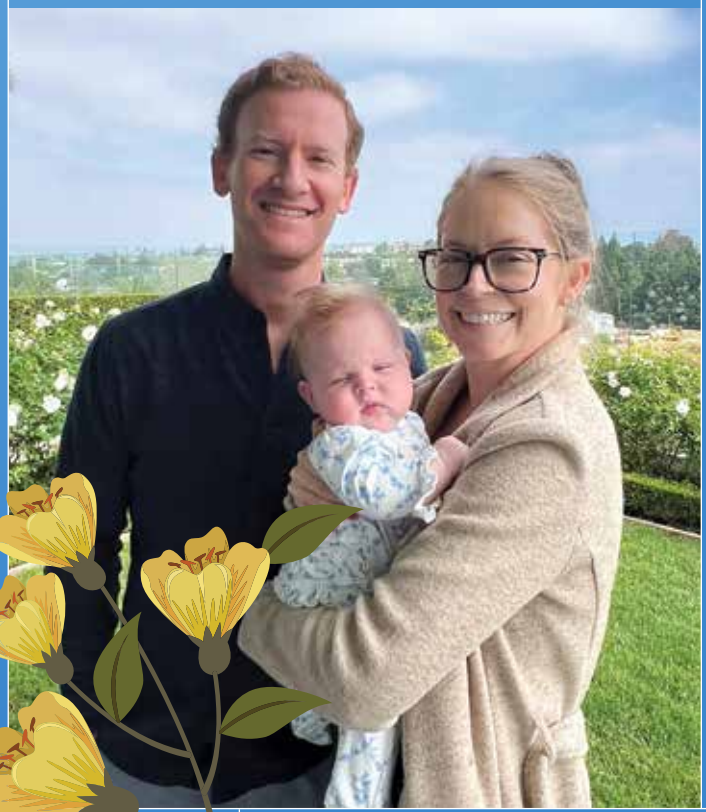
Fortunately, Danny selflessly decided to donate his kidney to the donor match program so that I can receive a kidney from a live donor. Not only will his kidney go to someone waiting desperately for this gift of new life, but Danny's gift will significantly shorten the time until I receive a new kidney which could otherwise have taken years.

In March, I celebrated my "third birthday" making it three years since my stem cell and gene therapy transplant in 2022. I continue to be on a very low dose of medication (cysteamine). I recently went to UCSD for my follow-up appointments and my results looked stable and my VCN increased!

Overall, I feel great, and I remain more hopeful than ever that the stem cell transplant will stop the progression of cystinosis. The stem cell transplant was a pivotal and life-changing event for me. I hope and wish that everyone with cystinosis will have the opportunity to have the stem cell transplant in the near future.

I am beyond grateful to the doctors, our community, and each one of you who not only helped save my life but who have given hope to others who have cystinosis. Thank you for never giving up on my wish to "have my disease go away forever."

Love, Natalie



CYSTINOSIS RESEARCH FOUNDATION

OUR STORY. OUR IMPACT.

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

Today, CRF is the largest private fund provider of cystinosis grants in the world and has funded 252 grants in 13 countries.

CRF has raised over $73 million, with 100% of your donations going to support cystinosis research. CRF is the driving force of cystinosis research that has directly resulted in advances in treatment including the FDA-approval of Procysbi® and a FDA-approved stem cell and gene therapy clinical trial. We have accomplished milestones and given hope to the cystinosis community that a better quality of life and a cure for cystinosis is possible.

CRF IS SYNONYMOUS WITH HOPE

cystinosis RESEARCH FOUNDATION

Charity Navigator FOUR-STAR 2025

Platinum Transparency 2025 Candid.

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. Buildup of cystine in the cells eventually destroys all major organs of the body, including the kidneys, liver, eyes, muscles, bone marrow, thyroid and brain. Medication is available to control some of the symptoms of this terrible disease, but cystinosis remains incurable. Cystinosis affects approximately 600 people, mostly children, in North America, and about 2,500 people 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.

CRF GLOBAL OUTREACH - 13 COUNTRIES

AUSTRALIA
BELGIUM
CANADA
FRANCE
GERMANY
INDIA
IRELAND
ISRAEL
ITALY
NETHERLANDS
NEW ZEALAND
SWITZERLAND
UNITED STATES

WE ARE GRATEFUL EVERY DAY FOR YOUR SUPPORT!

CRF's highly strategic approach to funding has resulted in two FDA approvals and several human clinical trials. The research dollars we have invested have been leveraged by over $25 million in grants from other funding agencies. Not only does CRF research help our community, but our discoveries are applied to more prevalent diseases and disorders. CRF-funded research has the potential to help millions of others.

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

100%
OF YOUR DONATIONS DIRECTLY SUPPORT CYSTINOSIS RESEARCH

SINCE 2003, CRF HAS:

RECEIVED
1 FDA-Approved Drug and
1 FDA-Approved Clinical Trial

1+1

FUNDED
252 Multi-Year Grants
in 13 Countries

252

PUBLISHED
115 Articles in Prestigious
Scientific Journals by
CRF-Funded Researchers

115

RAISED
More Than $73 Million
for Cystinosis Research

$73M

TOGETHER, WE ARE CHANGING THE COURSE OF CYSTINOSIS. THANK YOU!

IN SPRING 2025

CRF Awarded **4** New Research Grants

Totaling More Than **$559,000**

and Published **2** Articles in Prestigious Journals by CRF Researchers

The Proof is in Our Journey

In 2003, at the start of CRF, we funded our first two studies. Year after year, we awarded additional grants to brilliant researchers around the world. CRF's impact is profound – we have created a collaborative research community dedicated to unlocking the mysteries of cystinosis.

Today, in 2025, CRF has proven itself to be the global leader for cystinosis research, funding hundreds of researchers who are working tirelessly to find better treatments and a cure for cystinosis.

Hundreds of researchers are working on this rare disease because of CRF funding.

24/7 Research

CRF is funding researchers in 13 countries. This means someone is working every minute of the day to find better treatments and a cure.

We're Cystinosis-Specific

CRF funds research that targets every area of the body affected by cystinosis including the kidney, eyes, brain, bone and muscle.

And now more common diseases are beginning to benefit from CRF-funded research.

Meaningful and Lifelong Friendships

Hundreds of families connecting at the annual Day of Hope family conference. The CRF family grows every year and includes families from all over the world, confirming CRF's reach and commitment to everyone in the cystinosis community.

Over $73 Million

raised for cystinosis research bringing hope to the community.

FDA Approval

In 2013, Procysbi®, a delayed-release medication was FDA approved. CRF funded every early bench and clinical trial.

In 2018, the FDA approved the first stem cell and gene therapy treatment in six adults with cystinosis. The study was led by Stéphanie Cherqui, PhD, at UC San Diego.

Clinical Trial Advancements

As a result of the successful adult stem cell and gene therapy trial at UC San Diego, the next—and we hope the final—phase of the trial has recently commenced. This phase, led by Novartis, will treat young patients.

Gaining Global Exposure

115 CRF-supported, cystinosis-related published research papers to date.

Understanding Every Aspect of Cystinosis

25 clinical studies leading to new discoveries and treatments.
Since 2003, 252 research studies including critical animal models that CRF has helped create.

Biennial International Research Symposium bringing CRF-funded researchers together to accelerate research and establish collaborations.

First Canadian family conference hosted by CRF in 2022.

Establishment of the CRF Scientific Review Board comprised of the world's leading experts on cystinosis who guide the CRF research program.

Critical seed money provided to researchers in their early stages has been leveraged by other funding sources including NIH and CIRM.

WE'RE BETTER TOGETHER

Save the Date!

day of hope 26

CYSTINOSIS RESEARCH FOUNDATION
Family Conference

Mark your calendars now:

April 16-18, 2026

Huntington Beach, California

Together Again

We look forward to coming together once again with friends, families, and researchers to celebrate the CRF community and to strengthen the bonds that define our community.

Please save the date and plan to join us for a weekend filled with inspiration, discovery, and connection as we share the remarkable research progress we have made.

The conference offers a unique opportunity to hear directly from leading clinicians and researchers dedicated to improving the lives of our children and adults with cystinosis. This event not only rekindles our sense of hope but also keeps us informed about significant advancements in treatment options. Conference presenters will speak on a wide range of topics, including stem cell transplant therapy for cystinosis, kidney disease, bone and muscle disease, eye health, GI issues, mental health, impacts of diet, and more!

DON'T DELAY, REGISTER TODAY

www.eventcreate.com/e/2026-day-of-hope

Join us for inspiring research updates, powerful stories from our community, and a weekend of friendship and hope that brings us all together.

Cure Cystinosis International Registry

By Clay Emerson, PhD, PE, CFM

Brooke Emerson's dad, CRF Trustee, and CCIR Committee Member

Hammonton, New Jersey, USA

Cure Cystinosis
International Registry

With the assistance of researchers and physicians, the Cystinosis Research Foundation launched the new Cure Cystinosis International Registry (CCIR) in 2021.

The CCIR was designed to capture the unique experience of living with cystinosis, including the wide variety of necessary medications, kidney transplant, diagnosis, quality of life, and everything in between. Patients and caregivers from across the country have stepped up and contributed to the registry. The CCIR has lived up to its name with the international cystinosis community being represented by participants from 15 countries sharing their experience.

What's a "Registry" anyway?

Unlike the registries that we may be most familiar with, there are no countertop kitchen appliances, cookware, or bedding and bath linens involved. Instead, this registry provides a vital link between patients and cystinosis researchers. Due to the ultra-rare nature of the disease and the wide diversity of complications the disease presents, a patient registry provides the critically important link between patients and researchers. The reality with an ultra-rare disease is that progress depends on this link between the patient experience and the research community. Progress towards improved treatments and an eventual cure for cystinosis is only possible with input from our small patient community.

What can you do?

In order to better document the needs of the community and ultimately accelerate research, patients and caregivers can sign up for the registry. It only takes about 40 minutes to complete the questionnaire and be part of advancing progress. If you are a patient or caregiver that has already signed up, you are encouraged to update your responses to reflect current care, medications and complications. Existing participants will receive an annual reminder to update. The email will come with the subject "CoRDS Registry - Annual Questionnaire Reminder" and come from *cords@sanfordhealth.org*.

Sign Me Up

The CCIR Starter Guide

- WHO CAN PARTICIPATE?
  Cystinosis patients and caregivers (That's you!)
- HOW LONG DOES IT TAKE?
  40 minutes (That's it?)
- WHAT'S THE CATCH?
  No catch, just cool points. You're contributing to the progress our researchers are conducting to further advance a cure for cystinosis and related symptoms (That's wonderful!)

Please visit the CRF website to sign up today!

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

WINTER 2025

FAMILY & COMMUNITY UPDATES

FAMILY STORIES

COMMUNITY FUNDRAISING

MAKING OUR JOURNEY TOGETHER WITH HOPE

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.

Tuesday, December 2, 2025

GIVING TUESDAY – FUND A CURE FOR CYSTINOSIS! GIFT CHALLENGE
Cystinosis Research Foundation
www.cystinosisresearch.org

Saturday, December 13, 2025

THIRD ANNUAL PICKLEBALL TOURNAMENT IN HONOR OF ISLA MCALLISTER
LifeTime Fitness, Oklahoma City
Contact Duncan McAllister: duncanmcal@yahoo.com

Saturday, February 28, 2026

RARE DISEASE DAY
Cystinosis Research Foundation
www.cystinosisresearch.org

Spring 2026

ANNUAL FISHING FOR BROOKE'S CURE IN HONOR OF BROOKE EMERSON
Hope for Brooke
Contact Clay Emerson: clay.emerson@gmail.com

Month of April 2026

NATALIE'S WISH CELEBRATION ONLINE FUNDRAISER
Cystinosis Research Foundation
Contact Nancy Stack: nstack@cystinosisresearch.org

Thursday, April 16 – Saturday, April 18, 2026

CRF DAY OF HOPE FAMILY CONFERENCE
Hyatt Regency Hotel, Huntington Beach, California
Contact: jemerson@cystinosisresearch.org

Wednesday, October 21, 2026

SETH'S CIRCLE OF HOPE IN HONOR OF SETH DEBRUYN
Calgary, AB, CAN
Contact Kristen Murray: murraykristen@hotmail.com

NATIONAL KIDNEY REGISTRY®
FACILITATING LIVING DONOR TRANSPLANTS

What Is the National Kidney Registry (NKR)?

Imagine a kidney swap system—but on a national scale.

- When someone wants to donate a kidney to a loved one but isn't a match or they prefer more flexibility in timing, they can donate to a stranger through the NKR.
- In return, their loved one gets a "voucher" for a living kidney from a pool of compatible donors in the network.
- This creates donor chains: one kidney donation can set off a ripple of transplants across the country, saving multiple lives.

How the NKR Finds the Best Match

Eplet Matching: Precision in Compatibility

For decades, kidney transplants have been coordinated through basic blood type matching. We learned over the course of this year that the National Kidney Registry uses advanced software to analyze HLA proteins on kidney cells at the microscopic level.

- A low eplet mismatch between donor and recipient means the immune system sees the new kidney as "less foreign," which lowers the risk of rejection.
- The NKR uses this data to prioritize donor kidneys that are immunologically closest to the recipient — especially important for young patients who need their kidney to last decades.

Recently, Jenna Partington received a kidney with a very low eplet mismatch, which meant her Stanford transplant team was able to give her a steroid-free (prednisone-free) anti-rejection regimen, reducing side effects and protecting her long-term health.

In short: low mismatch means fewer medications and fewer complications.

Read about Jenna and Patrick Partington's experience with NKR on page 16.

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

Seventeen months with Jenna

Six surgeries. Ten months of dialysis. A sparkling new kidney. Jenna is going back to school!

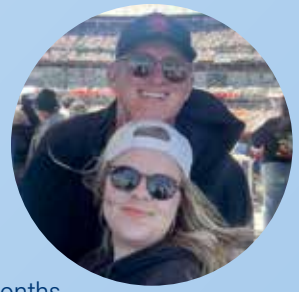
A TIMELINE OF MILESTONES AND MEMORIES

May 21, 2024

Milestone: Jenna arrives home from college to have her final knee surgery at Shriners, removing metal hardware placed during her osteotomy in 2019. She takes medical leave from the University of Idaho to prepare for a kidney transplant.
Memory: Jenna DJ'd as we listened to Midland and Niall Horan in the car during trips to appointments. We made plans for "carrots" on the horizon, to get us from one enormous undertaking to the next. Sometimes a meal, sometimes a series to binge on Netflix, sometimes a quick getaway in Northern California.

Dec. 20, 2024

Milestone 1: Kevin donates a kidney to Jenna at UC Davis Med Center.
Milestone 2: Jenna has two kidneys removed and receives Kevin's kidney.
Memory: Merry Christmas to all! What a beautiful thing to watch Kevin selflessly do what had to be done to help Jenna live and thrive. After surgery, we waited to see urine from the foley catheter. There was none. Jenna's stomach was bloated and her blood pressure was low.

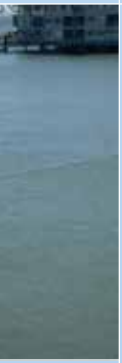


Dec. 21, 2024

Low Point: Kevin's kidney is emergently removed due to renal vein thrombosis. The transplanted kidney never got blood flow or made urine.
Memory: So many questions. Kevin was angry and heartsick. He wondered "Why didn't everyone's prayers work?" I suggested maybe they did, in that our daughter is alive after so much trauma over 48 hours.

Dec. 22, 2024

Milestone: Jenna begins dialysis three times a week for three hours each session for 10 months.
Memory: Anxiety. The dialysis center. The machines. The blood running out of Jenna's chest port and through thick tubes was a lot to take in. Fellow patients were much older than she, and had various critical health problems, which was also a lot to take in. Jenna had a few anxiety attacks in the beginning, and for a couple months my mom or I would sit and rub her feet to help her stay calm. In time, she offered smiles and encouragement to fellow patients, brightening their days. Most of them would WISH for only ten months of dialysis. She started going to treatments alone, indulging in three hours at a time of her new passion: watching new and old Formula One car races.



Jenna with Jeff Earle

Cousin Norm and Ellen

Apr. 24, 2025 **Milestone:** Friend and neighbor Jeff Earle donated a kidney through the National Kidney Registry (NKR), earning Jenna a voucher for a future transplant.
Memory: How do you thank a dear friend, golf partner, travel companion, stock broker and next door neighbor for an ORGAN? You simply can't. Jenna and Jeff have an unbreakable bond for life. We learned that Jeff's kidney was transported to UC San Francisco immediately following his surgery, and gifted to a woman from Gilroy, CA. The grateful recipient wrote a letter of thanks to Jeff that would bring anyone to their knees. Jeff framed the letter.

May 19, 2025 **Milestone:** Cousin Norm, who is in his early 70s and in stellar health, donates a kidney through the NKR, earning Patrick a voucher for a future transplant.
Memory: Norm, who until this year, we visited primarily at our family beach in McCall, traveled twice from Caldwell, Idaho, to Sacramento with his wife Ellen (once for evaluation, and once for kidney donation) in an act of incredible caring and generosity. Norm reminds me of my dad, and his presence and donation on Patrick's behalf, during a rather difficult season with Jenna, was very special. We shared walks and meals and a trip to Napa. Norm has been a shining example of what family does for family. What a blessing he and Ellen are!

Aug. 19, 2025 **Milestone:** Jenna received a very well matched living donor kidney through the NKR at Stanford.
Memory: We arranged a place to stay in Palo Alto to be near Stanford Hospital and Jenna's follow ups for a few weeks. (Kevin and I took some great hikes when we could break away!) At 7pm on transplant day, a kidney destined for Jenna arrived on a commercial flight from the East Coast. An Igloo cooler labeled "HUMAN ORGAN, HANDLE WITH CARE" (it has a GPS tracker inside, thank you very much) comes from an unknown person on the East Coast whose surgery was coordinated with Jenna's. Once transplanted at 9:30pm 8/19/25, it begins making urine immediately! Pure joy.

Oct. 1, 2025 **Milestone:** Next week, Jenna plans to continue her life at the University of Idaho. She is taking an online class, looking for a job, and preparing for what we hope will be the healthiest semester of learning in her life in Spring 2026!
Memory: I'm so happy for her! I'll sure miss the music filled car rides together, deli sandwiches, binging shows and simply being needed at a time when many mothers' nests are empty! Jenna has grown in spirit and strength these past 17 months. It's been emotional and trying, but I'll treasure the extra bit of time we've been able to spend with her.

TBD **Milestone:** Last week, we received two letters from Stanford Hospital on the same day. One was Jenna's final billing statement, and one was Patrick's appointment notification for his transplant consultation.
Memory: I thought, "Let's run it back and do this one more time!" To see Jenna with a sparkle in her eyes and flush in her cheeks is to want the same for Patrick, sooner than later!

"HUMAN ORGAN, HANDLE WITH CARE"

NATIONAL KIDNEY REGISTRY®
FACILITATING LIVING DONOR TRANSPLANTS

Learn more about the National Kidney Registry on page 15.

> Being an active part of this community, we have followed many stories of other kiddos just like her.

SUMMER 2025

ADVENTURE ✓
COMMUNITY ✓
HYDRATION ✓

Summer is finally winding down in Northwest Indiana (late September) and what a great summer we have had. Our fun kicked off with a few days of sunshine in April at the CRF conference in Huntington Beach, followed by a day at Disneyland in the most perfect weather. The location and facilities were incredible for this year's conference. Our whole family looks forward to it but for a few years now, Josie has remembered so many friends and cannot wait to connect again. Her brother, Brendan, has also made connections throughout the years and had a blast with the teens. With Josie being eight now, she asks a lot more questions and pays more attention. She also couldn't wait to see the little ones and play with them in the daily kids camp. These connections mean the world to all of us, and we are so grateful to the Stacks and all the planners for their dedication to this yearly event.

Our fun in the sun continued all over the Midwest after April, with school ending mid-May. Josie loved summer camp near home with her best friend and cousin Ella, vacation in Lake Geneva, WI, visiting friends in Illinois, beaches in Michigan, and, of course, swimming in our pool here at home. We also were able to take a road trip to meet new cystinosis families at a meetup in Grand Rapids, Michigan.

Installing our pool was a must a few years back as the heat started to show its effects on her health. It seems as soon as May hits, the air conditioning comes on and the pool is ready. Thankfully, Tom and I like to host, and Tom keeps the pool crystal clear, so we have become quite the party house.

Josie has now understood the importance of speaking up for herself and has become a great advocate for her needs. Trying not to bring cystinosis up constantly and just letting her be a kid in the summer does have its challenges. Stopping her from swimming to jump out of the pool for meds, showing up at camp or play dates early has her irritated but she understands. She knows she doesn't feel well outside for long being in the direct heat and needs to play inside or get into the water.

She attended summer camp during the day locally and thankfully, the counselors were helpful, keeping an extra eye on her and her massive water bottle. She has made close friends at school and their parents have been kind about her needs at play dates. As her fourth year at her current school has recently started, we have been lucky to have compassionate and understanding teachers, staff and a nurse who takes great care of her daily.

An hour of mini golf in mid-morning July with mom & dad, despite two bottles of water, hat and sunnies, saw her overheated and us both scared. It takes time to cool down and of course, there is always the worry about her kidney function. Leaving the home for an adventure out takes planning and lots of extra hydration. She has been stable for the past few years with her kidney function in the 40s. Being an active part of this community, we have followed many stories of other kiddos just like her. We know what her future will hold regarding her kidneys and hope to hold off on the kidney transplant for as long as possible. For now, we are and have been taking it daily, not sweating the small stuff and having as much fun as possible.

By Monica Gielarowski, Kaleb's maternal grandmother CHARLESTON, SOUTH CAROLINA

STRENGTH TO NEVER GIVE UP!

Kaleb turned 12 this past August. He still loves swimming, cartoons, video games and creating his own media. We often wonder what the future holds for him, especially with the medical challenges he faces every day. He says he just wants to be normal like other kids and enjoy life. He is in 5th grade this year and between in-person classroom and his school's homebound program, he is doing well.

Just three years ago, after suffering his first ever seizure, he was diagnosed with a glioneuronal brain tumor. While benign, it is intertwined with his optic nerve and remains inoperable. After 70 weeks of chemotherapy, he continues to be monitored at MUSC Children's Hospital having MRIs every three months, with stable results to date. The year before, he was diagnosed with IBD/ulcerative colitis. We often say and as crazy as it sounds... "if the only thing we had to worry about was cystinosis, it would be so much better."

HE FIGHTS A BATTLE NONE OF US CAN RELATE TO, AND ALONG WITH HIS STRONG SPIRIT HE TAKES ON WHATEVER IS THROWN AT HIM.

His doctors refer to him as "medically complex," so many times we just cannot get answers. We do believe there is a connection between cystinosis and his other conditions and bring it up with his specialists every chance we get. We advocate for awareness, providing his doctors with published research papers and/or new updates from CRF and the cystinosis community.

We continue to unite as a family in supporting his overall development. His mom and dad will say that it is a struggle at times, and the worry is 24/7. While at the same time, never giving up the search for answers in every aspect of his well-being. Special needs families often refer to the loneliness associated with living a different life and we strive to give him as much childhood normalcy as possible. He is blessed to have family that is involved and that strives to learn more about his disease.

Kaleb loves when he can get together with his aunts, uncles, and cousins from out of town. His paternal grandmother visits often, relaying that she sees him doing great things in the future. My husband and I live close by and help however we can. His grandpa says he is amazing and spends time with him often.

My daughter, Kate, is nothing short of amazing. She has become a true researcher, pushing for input on lab results, side effects and asking the hard questions needed for him to feel better. She and her husband, Mike, are true advocates for Kaleb in every sense of the word. Mike is Kaleb's best friend. Kaleb loves their time together in the evenings and on weekends. When Kaleb is feeling his best, we celebrate every moment. He fights a battle none of us can relate to, and along with his strong spirit he takes on whatever is thrown at him.

In the fall of 2022 after his brain surgery, Kaleb's Great Uncle Len presented him with a set of Naval Aviation Wings to recognize and honor his achievements. His heartfelt note below summed it up best and bears repeating for all cystinosis warriors. Kaleb is part of an elite group, along with the many other cystinosis children and young adults. You are all fighters, remain strong and never give up!

To Kaleb:

"Navy wings are given to Naval and Marine Aviators including many astronauts and those from Top Gun. They are worn to show everyone how hard they have worked and studied and how much they have accomplished. You are a very special person and have worked so hard and accomplished so much that I wanted you to wear these wings as a symbol of your great accomplishments. Like many military men and women who have been in battle, you fight and never give up. You are strong and stand up to others when you know you are right and accept what is given you when you know it is for the best. When you look at these wings remember you now represent an elite group of people who must remain strong. Never let anyone get you down. You will always be my guiding pilot and shining light."

Awarded by:
Leonard Deal, CAPT USN (Retired) – Nov. 2022

By Bianca Provost and Jonathan Goulet, Zoé's parents LEDUC, ALBERTA, CANADA

THE GOULET FAMILY

SEEN THROUGH THE EYES OF 4-YEAR-OLD NOAH

This is our story from the eyes of our son Noah, regarding his little sister who has cystinosis. As parents, we strive to include everyone in this process, ensuring each person feels valued, supported, and a true sense of belonging.

Last summer, when I got home from daycare with my dad, I began searching the house for my mom and Zoé, but daddy told me they left for the hospital because Zoé was feeling sick. That's where our story all started!

BONJOUR!

My name is Noah and I'm Zoé's big brother. Zoé is two years old now. She is funny, smart, and fearless. She likes to dance, splash in the water, watch butterflies, jump on the trampoline, and copy everything I do. When Zoé was born, my parents told me that my job as her big brother was to always help and support her. Sometimes, Zoé has a hard time eating so I have to show her how it's done. I remind her how to hold her spoon and encourage her whenever she takes a bite.

One day, my parents told me that Zoé was diagnosed with something called cystinosis. I never heard that word before! I learned that cystinosis makes her body work differently and that mom and dad are often busy helping Zoé stay healthy. Zoé has to take a lot of vitamins every day with long weird names. Mom said that it makes Zoé unique and precious, just like I am.

Mom and Zoé go to the hospital more often than other kids. I don't always understand why but I enjoy going to see them and meeting the doctors. I also get to have "boys' nights" with dad so I don't miss mom and Zoé too much.

Zoé has a busy schedule, and I like to help my family. I like to push food into Zoé's G-tube, make water syringes, put a towel under her chair when she feels nauseous at dinner and make her laugh when she is sad. I also give the best hugs to help Zoé process her emotions at daycare. I do everything I can to make Zoé feel better because when Zoé is too sad or upset, she can throw up.

Our family has changed a lot since Zoé was diagnosed but we have also gotten closer to one another. At the beginning, my mom and Zoé were in the hospital for five long weeks. **That's when they put a "button" into my sister's belly. It's like a magic door that brings food and vitamins directly into her stomach without her having to taste it!**

Change is difficult and a lot of things have changed in our house since Zoé was diagnosed, but the good news is that everyone is feeling better now. Mom now spends

more time on self-care and says it helps her to be a better mother. Dad enjoys golfing and fixing things around the house. Zoé is finally growing, gaining weight, and getting stronger. My parents even put me in play therapy for a short time where I got to talk to someone and play with her cool toys. My parents wanted me to have a safe and comfortable place to share my thoughts and feelings.

You know what I love the most about cystinosis? I love to go see Zoé's kidney doctor in Calgary. We get to stay in a hotel with a pool, and Dr. Midgley is very funny. I also enjoyed going to the CRF "Day of Hope" Family Conference because I meet so many friends and California doesn't have snow in April! My favorite part was eating all the desserts and playing with my new friends! I think that day made my parents happy and feel better. We also, as a family, started to support CRF by making a Canada Helps page for Zoé, and organizing some events to collect donations for the foundation! We had a bottle drive, a 50/50 raffle, and a silent auction in honor of Zoé. And we will always celebrate July 9 with a cake and positive thoughts. We also encouraged our friends and family to light a candle on this day to bring light and joy on the day my sister was diagnosed. My mom was inspired by another family who did this, and thought it was a wonderful idea.

Even though cystinosis can be complicated, I think Zoé is very brave and strong. She never gives up, and she is always smiling.

I don't know what the future will look like, but I do know that Zoé and I will keep playing, laughing, and learning together. And no matter what, I'll always be right here by her side.

—Her big brother and her best friend,

Noah Goulet

We light a candle on July 9 to bring light and joy on the day my sister was diagnosed.

By Shelly Suetta, Emma's mom ETNA, CALIFORNIA

"Everything happens for a reason!"

This is a quote that I have lived by. It has been nine years since Emma spent a month in UC San Francisco children's hospital. This hospital is six hours from our home. Our lives were turned upside down on June 17, 2015. This is the day that we got the official diagnosis of cystinosis. Within a week, I was connected to the very small community of other cystinosis families. We are so thankful for social media. I'll never forget the call I got from Zeke to walk me through life with this new diagnosis and how to move forward. His daughter, Zylar, was a few years older than Emma, so he had already walked in our shoes. We talked about medications and how to avoid making Emma sick. We discussed the best foods to feed her to keep her tummy happy. Since that day, we have become a part of a new "cysta family." Zeke, Taishia, and their family drove 12 hours to come and stay with us in 2017. Ever since then, we have gone to visit their family multiple times and made many trips to Disneyland together.

"Every cloud has a silver lining!" We attended our first "Day of Hope" conference in the spring after Emma's diagnosis. It was overwhelming and heart wrenching, but amazing and welcoming at the same time. We met our new forever friends and family that weekend. We met Emma and Gracie, twins with cystinosis, connected to my Emma Grace by name and diagnosis. We also met Jenna and Patrick, whose mom, Teresa, visited us at UCSF during one of our many hospital stays. Emma has built lifelong friends whom she calls her "cystas". She met her best friend Emma D. two years ago at the "Day of Hope." Since they met, there are very few days that go by that the two Emmas don't talk on video chat and play together. They are already planning our next trip to the "Day of Hope" and all the possible ways they can be together, including a hopeful sleepover.

My oldest daughter, Lillyanna, has also made deep connections with friends and siblings of children with cystinosis. She talks daily to her best friend, Peytan, who is 14 with cystinosis. They have not yet met in person but had an instant connection because of cystinosis. I think it helps her to understand her sister's struggles a bit more, having friends that are going through the same disease. She looks forward to the "Day of Hope" just as much as the rest of us because she gets to see all her friends that she would never have met without her sister's diagnosis.

"My life is perfect, even with cystinosis!" Emma has said this to me a couple times this past year. Recently, we have talked about the possibility of a "Make a Wish" for Emma in the future. When I asked Emma what her wish might be, she responded, "Nothing, my life is perfect, and I don't need to change anything." She knows all about her medications and knows how to do them all via her G-tube. She doesn't yet eat by mouth but plans to be a chef after attending culinary school. She knows more about food and cooking than most adults she talks with. It makes my heart so happy to know she is so happy with her life despite her struggles.

Nine years ago, our hearts were broken with our new diagnosis and for all the changes our lives had. The round-the-clock care, doctors' appointments, blood draws, blended food, therapy, medication, pharmacies and so much more were unimaginable. Since then, our lives have found a new normal. We have made lifelong friendships that we consider to be family. We have found so many silver linings to be thankful for. There would be so many things we would not have, and would not have the opportunity to do without cystinosis.

When Friendships Turn Into Family

TOGETHER, WE ARE one

1 PURPOSE. 1 JOURNEY. 1 CURE.

COMMUNITY FUNDRAISING

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

The Tschannen Family – Brookfield, Missouri

8TH ANNUAL WESTON TSCHANNEN MEMORIAL GOLF TOURNAMENT

The Tschannen family proudly hosted the 8th Annual Weston Tschannen Memorial Golf Tournament on Saturday, May 17, at the Brookfield Country Club. The event brought together enthusiastic golfers and generous sponsors, raising more than $17,000 for the Cystinosis Research Foundation (CRF) and local organizations close to Weston's heart. It was a beautiful day to enjoy golf while honoring Weston's memory, made even more meaningful by the strong support of friends and the community. Since the first tournament in 2017, the event has raised more than $76,777 to advance life-changing cystinosis research. CRF is profoundly grateful for the Tschannen family's steadfast dedication and for everyone who makes this tournament a success year after year. Together, we are bringing hope and progress to children and adults with cystinosis.

The Fehr Family – Saskatchewan, Canada

HOPE FOR JAMES GOLF TOURNAMENT

The Hope for James Golf Tournament took place on Saturday, August 9, 2025, at the Rosthern Valley Regional Park Golf Course in Saskatchewan, Canada. It was a beautiful day with perfect weather as 27 teams (a record turnout of 108 golfers!) participated to honor James Fehr and support cystinosis research. Each year, family and friends in the community make significant contributions by playing golf, purchasing sponsorships, bidding on silent auction items, and buying raffle tickets. This year, the tournament raised more than $15,000 through Canadian partnerships and Canada Helps.

We extend our heartfelt thanks to the Fehr family and all the participants who helped make the tournament a tremendous success. We appreciate our Canadian families and friends who support the Cystinosis Research Foundation and our mission to fund research for improved treatments and a cure for cystinosis!

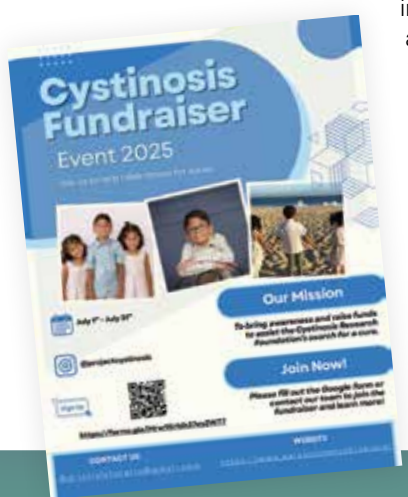
RAISED
MORE THAN
$15,000

The Khalasi Family – Sacramento, California

"DRY IN JULY" FUNDRAISER, IN HONOR OF AARAV KHALASI

The "Dry in July" Cystinosis Fundraiser was a student-led initiative organized by a dedicated group from Sacramento Country Day High School, determined to make a difference in the fight against rare diseases. Inspired by the resilience of Aarav Khalasi – who bravely manages daily medications and the challenges of living with cystinosis, a rare and life-threatening genetic disorder – classmates, led by Kale Patel, came together as a marketing team to raise awareness for this devastating condition.

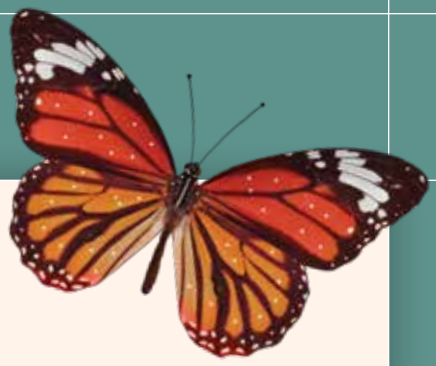
In partnership with Aarav's Time to Shine and the Cystinosis Research Foundation, the campaign invited the community to take part in a simple yet powerful challenge: to refrain from adult beverages for the month of July to support vital research. Whether giving up the beverages yourself or sponsoring someone who did, participants made a remarkable impact, raising over $6,515 for cystinosis research.



We are deeply grateful to Kale and his team, Ava Levermore, Itzel Zamora, and Rebecca Lin, for their creativity and commitment. On behalf of CRF, the Khalasi family, and the cystinosis community, we would like to thank you for your efforts in bringing hope and a brighter future to those living with cystinosis.

RAISED
OVER
$6,515

TOGETHER, WE ARE one



Cystinosis Research Foundation – Irvine, California

Every April, we celebrate another year of groundbreaking cystinosis research!

Last year's Natalie's Wish Fundraising event raised over $1 million for cystinosis research! Every dollar brings us closer to a cure and more effective treatments. Your generosity fuels life-changing discoveries and gives hope to those affected by cystinosis.

It all began with one girl's wish "to have my disease go away forever." More than two decades later, Natalie's dream has become a global movement to find a cure. Today, that cure feels within reach — thanks to your commitment to children and adults with cystinosis.

From a few families to a united worldwide community, our shared determination continues to drive progress. Much has been achieved, but there's still more to discover.

Natalie's wish is our wish. Join us in celebrating 22 years of impact and continuing the momentum toward a cure for all born with cystinosis.

Natalie's Wish 26

THE WISH THAT IS CHANGING LIVES

The Galloway Family – Cumming, Georgia

COLLINS' CURE CUP GOLF TOURNAMENT & SILENT AUCTION

Collins' Cure

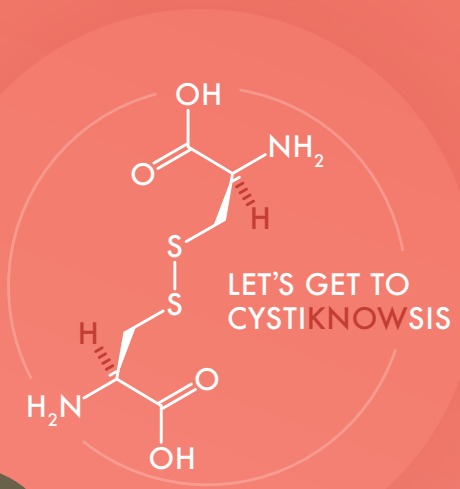
Christina, Hunt, Rowyn, and Collins Galloway are overwhelmed with the generosity and support they received for their 5th Annual Collins' Cure Golf Tournament and Silent Auction held on November 8, 2025. The fantastic response from their Cumming community has been inspiring, and their generosity ensures that Collins' Cure can continue supporting the Cystinosis Research Foundation, with all proceeds going directly to advance their vital work — funding research, supporting families, and driving progress toward better treatments and, one day, a cure.

Thanks to these collective efforts, they raised over $190,000, almost double what they brought in last year! The proceeds raised will play a crucial role in advancing research and treatment options and ultimately bringing us closer to finding a cure. The spirit of generosity and compassion displayed by their friends and community is inspiring. It helps us continue our mission and reinforces the power of community when we come together for a common cause. Your support goes beyond financial contributions; it symbolizes hope, unity, and the belief that positive change is indeed possible.

CRF is deeply grateful to the Galloway Family for their time and commitment in hosting their annual golf tournaments. We extend our heartfelt thanks to everyone who participated, donated, volunteered, and contributed in any way to the success of this event. Your generosity has a lasting impact on the cystinosis community by funding research for better treatments and a brighter future for everyone affected by this rare disease. Thank you!

WINTER 2025

RESEARCH UPDATES



OCTOBER 2025

The next step in gene therapy for cystinosis is underway!

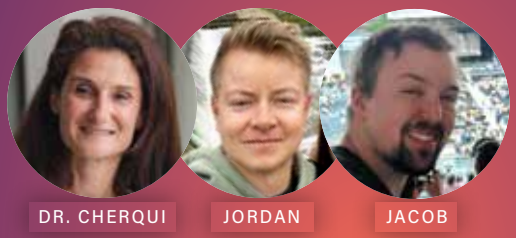
NOVARTIS' CYSTEM STUDY IS NOW OFFICIALLY RECRUITING PATIENTS.

BY **STEPHEN L. JENKINS, MD**

ASSISTANT PROFESSOR, HOSPITALIST, DEPARTMENT OF INTERNAL MEDICINE
UNIVERSITY OF UTAH HOSPITAL — SALT LAKE CITY, UTAH

THE BRAVE BEGINNING

We honor Dr. Stéphanie Cherqui's successful completion of the Phase I/II trial with adults: Jordan Janz (Oct. 7, 2019); Jacob Seachord (Nov. 16, 2020); Tyler Joynt (Nov. 15, 2021); Natalie Stack Morgan (Mar. 29, 2022); and Kurt Gillenberg (Oct. 24, 2022) who each received a gene-modified autologous stem cell transplant.



DR. CHERQUI | JORDAN | JACOB

TYLER | NATALIE | KURT

The long-awaited next step in gene therapy for cystinosis is underway. After the completion of the Phase I/II trial by Dr. Stéphanie Cherqui at University of California, San Diego, Novartis purchased the gene therapy program in 2023 and is now officially recruiting patients for the CYStem study.

In Dr. Cherqui's trial, six adults with cystinosis received a gene-modified autologous stem cell transplant. That means that the patient's own hematopoietic stem cells were harvested and the correct CTNS gene was loaded into those cells. After a brief course of chemotherapy to make room in the bone marrow, those genetically corrected cells were infused back into the patients. Fortunately, none of the brave participants in that trial suffered any major adverse effects from the gene therapy, demonstrating that the treatment is safe. Dr. Cherqui also showed a reduction in white blood cell cystine levels, and most of the patients were able to remain off cysteamine.

But is the gene therapy a cure? And is it safe for children? The next phase of the trial, which will be conducted by Novartis, will try to answer these questions. Novartis has many years of experience developing gene therapies and bringing them to patients. In 2019 the FDA approved a gene therapy for spinal muscular atrophy, and since that time, more than 4,000 patients have received it around the world.

To prove that a treatment works, you need to compare it to a control group. In the CYStem study, there will be two cohorts, and cohort 0 will be the comparison group. Cohort 0, led by Dr. Larry Greenbaum at Emory University, will continue on standard-of-care cysteamine therapy. Their outcomes will provide the baseline needed to measure the true impact of gene therapy.

Cohort 1 is the treatment arm and will receive gene therapy at either UC San Diego/Rady Children's Hospital or Stanford University. Novartis plans to enroll 15 participants, ages 2 to 5 years old with a diagnosis of cystinosis and evidence of Fanconi syndrome. The primary endpoint that Novartis is hoping to show is that the stem cell treatment reverses Fanconi syndrome. They will also collect patient-reported outcomes, like how patients feel and function after the treatment. This is a combined Phase I/II study, which means researchers are testing both safety and early signs of effectiveness. At this stage, not meeting the primary endpoint does not mean failure – it simply guides how the study evolves. It is normal to adapt endpoints based on the data that they collect during the study.

The study is recruiting 2 to 5-year-olds because Novartis wants to maximize the possibility of detecting a treatment effect, and this is the age group with the best chance. This does not mean that if the FDA later approves the drug, it will only be available to young children. By the time the product comes to market, the goal is for it to be available to everyone with cystinosis. After the first six patients are treated, the Data Monitoring Committee will review all the data to determine whether there is benefit, and whether the eligibility criteria can be expanded.

None of this would be possible without the Cystinosis Research Foundation community. Thanks to your support, Dr. Cherqui was able to bring this project from the laboratory bench to our patients, and Novartis is carrying it forward. Every lemonade stand, bake sale, golf tournament, and gala has led to this milestone. It has been a long journey, and we still have a long journey ahead, but together, fueled by hope and our support for one another, we move closer to a future where every person with cystinosis can live a healthier life.

THE GOAL IS TO FIND TREATMENT FOR EVERYONE WITH CYSTINOSIS

PATIENT BY PATIENT, PIECE BY PIECE

by Dennis Arp

WITH HER BIG-PICTURE APPROACH, **DR. AUDE SERVAIS** ADDS MOUNTAINS OF MUSCLE AND DECADES OF DEFINITION TO A GLOBAL RESEARCH VISION.

With each new day, the levers of cystinosis research grow stronger and more sophisticated. Perhaps no one contributes more to this steady drumbeat of progress than does Dr. Aude Servais, who uncovers countless insights at the cutting edge of exploration.

So, here's the irony. Despite the expansive reach of today's research tools, none can chart the apparently limitless depth of Dr. Servais' commitment to improving the lives of cystinosis patients.

For more than two decades, Aude Servais, MD, PhD, has bridged the worlds of cystinosis clinical treatment and research investigation. Even as she provides world class care to more than 30 adult cystinosis patients via her nephrology practice at Hospital Necker in Paris, she and her lab team also dive into a host of vital research realms, thanks largely to support from the Cystinosis Research Foundation (CRF) and its donors.

From the multi-organ effects of cystinosis, to the complications of the central nervous system; from cellular and molecular mechanisms, to renal transplantation; from cysteamine therapy, to swallowing dysfunction, Dr. Servais turns discovery into delivery – a true hero of the bench-to-bedside dynamic.

However, Dr. Servais deflects all talk of heroism and instead shines a light on the courage of cystinosis patients and their families.

"Everything we do is driven by the needs of those who live every day with the impact of this disease," she says. "This community is always at the center of our work."

UNEARTHING NEUROMUSCULAR INSIGHTS

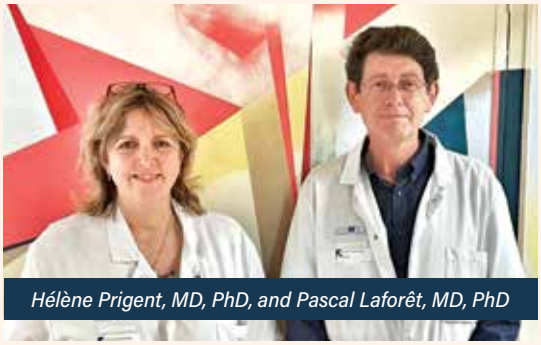
As her research work continues to diversify and deepen, a natural progression leads Dr. Servais and colleagues to want to learn more about neuromuscular complications. In the early days of her clinical life treating cystinosis, her patients were all young, so many had not lived long enough to experience a wide variety of neuromuscular complications. Now patients are living into adulthood, and those complications are presenting new challenges.

The good news is that better understanding, reliable diagnosis and early treatment offer paths to translating the team's research into real-world impact.

Dr. Servais' latest work targeting neuromuscular defects has reached a culminating phase full of insights that will inform next steps, including clinical applications.

The project began more than two years ago and is a multidisciplinary collaboration with co-investigators Pascal Laforêt, MD, PhD, and Hélène Prigent, MD, PhD. The research partners bring diverse expertise to the exploration of muscle function.

"We're all very excited because little is known about neuromuscular involvement with cystinosis," Dr. Laforêt said at the outset of the project.



Hélène Prigent, MD, PhD, and Pascal Laforêt, MD, PhD

SKELETAL PATTERNS EMERGE

Thanks to the participation of 20 cystinosis patients at the Raymond Poincaré Hospital Neuromuscular Center, more is now known about neuromuscular defects. Using high-level and standardized tools to assess muscle strength and function in each patient, the researchers are better able to describe patterns of skeletal muscle involvement.

In addition, respiratory muscle complications are becoming better understood, as are specific pulmonary functions and the consequences of cystinosis on breathing efficiency.

CYSTINOSIS RESEARCH STAR

Aude Servais, MD, PhD

Senior Nephrologist, Department of Adult Nephrology

NECKER HOSPITAL, PARIS DESCARTES UNIVERSITY

PARIS, FRANCE

The doctors are still parsing their data, but Dr. Servais is able to offer a range of insights gleaned from 30+ months of investigation made possible by CRF support.

All project participants are 18 or older and have significant histories with cystinosis, although not all have experienced neuromuscular defects or complications. They come from a wide range of backgrounds and experiences.

In addition to whole body Magnetic Resonance Imaging (MRI), all participants received a complete clinical assessment, including a respiratory exam to check lung function, a sleep evaluation, handgrip assessment, and tests of muscles used in swallowing. For every neuromuscular area, patients were given an assessment score.

"It's important to quantify effects muscle by muscle using standardized tests so we can correlate neuromuscular effects with the other complications of the disease," Dr. Servais says.

Some general takeaways have emerged:

- The muscles most affected by cystinosis are of the hand and the tongue. "It's particularly important to have this picture of the tongue because it plays such a critical role in swallowing," Dr. Servais notes.
- Some patients display weakness in the distal muscles of their legs, particularly in their lower legs and feet. Some also experience defects in their shoulder muscles.
- "We also found that patients who began treatment early in their lives tend to have far fewer defects across our neuromuscular evaluations," the doctor says. "We know now that early treatment is very important in reducing the long-term complications."
- There's a definable link between high cystine levels at the time of assessment and insufficient treatment until recently. In some cases, cystinosis may have been diagnosed early on and a treatment regimen prescribed, but patients struggled with compliance, Dr. Servais notes.

Whatever the scores from the evaluations, the goals of the overall research go beyond identifying neuromuscular risk factors and global patterns of vulnerability.

TRANSLATING FINDINGS TO MEET EVERYDAY NEEDS

"We want to be as patient-specific as possible because we're committed to improving the quality of life for each cystinosis patient we see," Dr. Servais says.

With that specificity in mind, patients in the study are getting help in adapting to their individual everyday needs.

"We now have a heat map of each muscle group, so we have a specific pattern, patient by patient," the doctor says. "The first step is to understand what's happening, because this is an understudied area of research, but then it's also important to begin implementing a therapeutic response."

As part of the study, participants received specialty care as needed. A team approach emerged and may provide a care model going forward. For instance, if a hand muscle defect is identified, the patient might begin a weekly program of exercises, or a specific tool might be used to prevent muscle retraction during sleep.

Some study participants received nocturnal ventilation to address a sleep-related defect, while others consulted with an ergotherapist to help with physical, emotional or developmental challenges posed by a neuromuscular complication.

"We are finding that this is a muscle disease very different from other muscle diseases," Dr. Servais says.

The doctors now know that cystinosis targets muscles through their vacuoles – the tightly bound membranes within cells that perform vital functions of health maintenance. What Dr. Servais and her colleagues don't know yet are other details of the disease's pathophysiology.

> > > >

www.cystinosisresearch.org 33

Muscle biopsies might be the next phase of her research, the doctor adds, "so we can study muscle function at the molecular level."

REVIEW BOARD ROLE FEEDS GLOBAL PERSPECTIVE

It's no surprise that even as she continues to analyze the results of her intensive multi-year study, Dr. Servais is already looking toward the next level of understanding. That commitment, stacked upon her decades of comprehensive cystinosis experience, makes her an invaluable member of the CRF Scientific Review Board.

As a board member, Dr. Servais collaborates with other experienced cystinosis scientists, researchers and clinicians from all over the world to evaluate every research application submitted to the foundation. Together, Dr. Servais and her colleagues have helped CRF fund 252 research projects in 13 different countries.

The Review Board role gives her a window to the vital function CRF continues to play in creating transformational change.

"It's rewarding to review proposals that reflect the range of research projects, from the fundamental ones dealing with the structure of cystinosin to those focusing more narrowly on the structure of treatments," Dr. Servais says. "I try to bring the clinical as well as the research perspective, to keep the work focused on patient needs."

She's quick to add that while fundamental research doesn't always lead directly to new treatments, it can open doors that over time yield extraordinary results. If CRF didn't fund such research, there's a good chance it wouldn't happen.

"Stem cell therapy is one of the most important achievements we've seen, and we know that the first study came from CRF funding," the doctor notes. "It's important to take such a long-term view while also supporting less ambitious projects that can lead to equally important advances in treatment."

OPENING DOORS TO NEW TREATMENTS

Identifying deserving projects is like adding pieces to a life-affirming puzzle, she says.

"In the end, we hope to improve our overall knowledge of the disease, and when we describe a new mechanism, we open the door to specific new treatments," Dr. Servais adds.

That expansive perspective brings her back to the importance of cross-disciplinary collaboration and the possibilities offered by her neuromuscular research.

With their project, she and her colleagues are adding another important piece to that global research puzzle, now decades in the making but more vital than ever.

"The first patients I saw had not been treated before, and we were seeing severe complications that were not well understood. Now I see older patients, and some have no complications – they are involved in important life projects with plans for even more," she says.

"Things have changed in wonderful ways," Dr. Servais notes with a pause for reflection. "We can see the future with much more optimism."

Stem cell therapy is one of the most important achievements we've seen, and we know that the first study came from CRF funding.

RECENTLY 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.

Genome-wide DNA Methylation Analysis Identifies Kidney Epigenetic Dysregulation in a Cystinosis Mouse Model, published in *Frontiers in Cell and Developmental Biology*

21 AUGUST 2025

by Giusi Prencipe, PhD

Laboratory of Rheumatology,
Ospedale Pediatrico Bambino Gesù
Rome, Italy

Nephropathic cystinosis is a rare genetic disorder characterized by cystine accumulation in lysosomes that causes early renal dysfunction and progressive chronic kidney disease. Although several metabolic pathways, including oxidative stress and inflammation, have been implicated in the progression of renal parenchyma damage, the precise mechanisms driving its progression are not fully understood. Recent studies suggest that epigenetic modifications, particularly DNA methylation (DNAm), play a critical role in the development of chronic kidney disease.

Clinical Perspectives on Myopathic Complications in Nephropathic Cystinosis, published in the *Journal of Rare Diseases*

JUNE 2025

by Reza Seyedsadjadi, MD

Department of Neurology,
Massachusetts General Hospital
Boston, Massachusetts

Dr. Reza Seyedsadjadi has been focused on research that addresses the devastating complications of myopathy and muscle wasting in cystinosis. This article provides a clinical perspective on the impact of myopathy in cystinosis and discusses strategies that will help identify, assess, manage, and mitigate these complications. Since 2019, the Cystinosis Research Foundation has been honored to support Dr. Seyedsadjadi's important work, awarding more than $826,908 in research grants.

To access the 2025 Published Studies, visit:
www.cystinosisresearch.org/published-studies

RESEARCH AND FELLOWSHIP

GRANT AWARDS

Cystinosis Research Foundation is the largest fund provider of grants for cystinosis research in the world, issuing 252 grants in 13 countries since 2003.

Sergio Catz, PhD *(Mentor)*
Mouad Ait Kbaich, PhD
(Fellow)
The Scripps Research Institute
La Jolla, California

"Cellular, Molecular, and Multi-Omics Characterization of Autophagy-ER Homeostasis Crosstalk in Cystinosis"

$150,000
TWO-YEAR FELLOWSHIP

Olivier Devuyst, MD, PhD
Principal Investigator
University of Zurich
Zurich, Switzerland
Francesco Emma, MD
Co-Principal Investigator
Bambino Gesù Children's Hospital
Rome, Italy

"Translaltional Value in SGLT2 Inhibitors in Cystinosis"

$296,900
TWO YEAR-STUDY

Umber Dube, MD, PhD
Principal Investigator
Stéphanie Cherqui, PhD
Co-Principal Investigator
University of California, San Diego

"Cutaneous Manifestations of Cystinosis as Biomarkers and Companion Diagnostics"

$109,204
TWO-YEAR STUDY

Jennifer Hollywood, PhD
University College Cork
Cork, Ireland

"Evaluation of a Novel Drug Combination Treatment of CF10 and Everolimus for Nephropathic Cystinosis in a New Cystinotic Rat Model"

$3,090
LAB EQUIPMENT GRANT

SEE LAY ABSTRACTS STARTING ON NEXT PAGE

Cellular, Molecular, and Multi-Omics Characterization of Autophagy-ER Homeostasis Crosstalk in Cystinosis

Sergio Catz, PhD, *Mentor*
Mouad Ait Kbaich, PhD, *Fellow*
THE SCRIPPS RESEARCH INSTITUTE, LA JOLLA, CALIFORNIA

OBJECTIVE/RATIONALE:

In cystinosis, defective cellular waste removal systems (autophagy pathways) contribute to progressive kidney damage despite current treatments. Our preliminary data indicate that increasing levels of a trafficking protein, Rab11-FIP4, can restore these cellular recycling mechanisms and reduce cellular stress markers. This project will define the molecular mechanisms underlying this restoration, providing evidence for developing complementary therapies that may help preserve kidney function in cystinosis patients.

PROJECT DESCRIPTION:

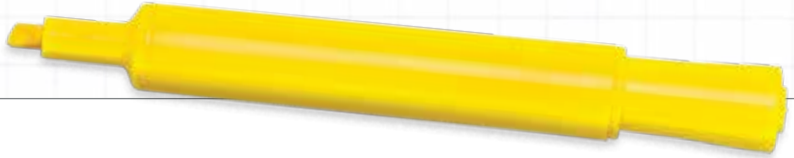
This study will examine three interconnected cellular recycling pathways (chaperone-mediated autophagy and macroautophagy) that show impairment in cystinotic cells and also explore other affected recycling pathways such as ER-phagy. Using patient-derived kidney cells, we will characterize defects in these pathways and test whether Rab11-FIP4 reconstitution corrects them. Our methods include super-resolution microscopy to visualize protein complex formation, comprehensive gene and protein expression analysis to identify affected molecular networks, and functional assays measuring kidney cell activities including protein reabsorption and transporter function. We will investigate interactions between Rab11-FIP4 and regulatory proteins NEDD4 and Rab24 and assess whether this intervention improves cell survival under stress conditions and potentially modifies the current cysteamine requirements.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

This research addresses cellular dysfunction mechanisms that persist despite cysteamine therapy. By elucidating how Rab11-FIP4 restores cellular recycling pathways, we aim to identify targetable mechanisms for therapeutic intervention. These findings may inform development of supplemental treatments that, combined with current therapy, could better preserve kidney function and potentially delay progression to end-stage renal disease.

ANTICIPATED OUTCOME:

We anticipate identifying specific molecular targets within cellular recycling pathways that are normalized by Rab11-FIP4 intervention. The study should yield: validated biomarkers for monitoring cellular dysfunction and treatment response; characterization of 3-4 key regulatory proteins suitable for therapeutic targeting; and mechanistic understanding of how trafficking proteins influence cellular stress responses in cystinosis. These findings will provide a foundation for developing targeted therapies.



Translational Value in SGLT2 Inhibitors in Cystinosis

Olivier Devuyst, MD, PhD, *Principal Investigator*
UNIVERSITY OF ZURICH, ZURICH, SWITZERLAND
Francesco Emma, MD, *Co-Principal Investigator*
BAMBINO GESÙ CHILDREN'S HOSPITAL, ROME, ITALY

OBJECTIVE/RATIONALE:

Cystinosis is a rare genetic disorder that causes cystine to accumulate in cells, damaging many organs, especially the kidneys. The most severe form disrupts kidney tubule function, leading to loss of vital substances and kidney failure. Current treatments have limits and side effects. SGLT2 inhibitors, which slow kidney disease progression in other conditions, may improve kidney tubule function in cystinosis. This project aims to explore these effects to develop safer, more effective treatments for children with cystinosis.

PROJECT DESCRIPTION:

This study will investigate how SGLT2 inhibitors protect kidney tubule cells in cystinosis. Using mouse and rat models, we will evaluate long-term effects and potential side effects. We will also design a phase 2 clinical trial protocol to test the safety and effects of dapagliflozin on children with cystinosis who have preserved kidney function. The ultimate goal is to prepare for a future clinical trial.

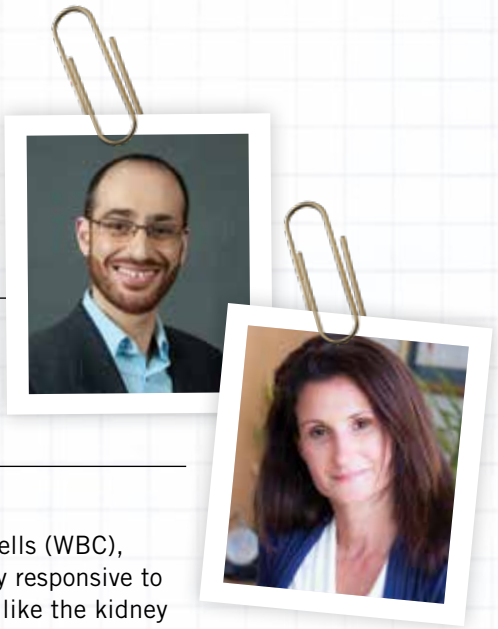
RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

This project will enhance understanding of cystinosis at the cellular level and help develop new therapies. SGLT2 inhibitors may help protect kidney and, in addition to cysteamine treatment, may slow the progression of kidney failure.

ANTICIPATED OUTCOME:

We expect to demonstrate that SGLT2 inhibitors reduce kidney damage and improve kidney function in preclinical cystinosis models even after evidence of tubular disease has developed. These data will be very important in the potential development of a clinical trial.

Advancing Non-Invasive Skin Cystine Crystal Imaging as a Biomarker and Companion Diagnostic for Cystinosis



Umber Dube, MD, PhD, *Principal Investigator*
Stéphanie Cherqui, PhD, *Co-Principal Investigator*
UNIVERSITY OF CALIFORNIA, SAN DIEGO

OBJECTIVE/RATIONALE:

Current cystinosis monitoring relies on measuring cystine in white blood cells (WBC), necessitating an invasive blood draw. However, WBC cystine levels are very responsive to cysteamine and do not fully reflect cystine crystal accumulation in organs like the kidney and skin. Non-invasive imaging of cystine crystals in the skin may provide a more accurate estimate of the body's total burden of cystine crystals. Here we propose to further develop and advance skin reflectance confocal microscopy as a reliable biomarker and companion diagnostic for cystinosis.

PROJECT DESCRIPTION:

We will analyze the world's largest dataset of skin reflectance confocal microscopy images in the context of cystinosis. This includes data from over 90 individuals with cystinosis and is expected to grow each year. First, using this unique resource which includes repeated scans from many of these individuals we will conduct a detailed longitudinal analysis to define the natural history of how skin cystine crystals progressively accumulate over time, even while patients are on systemic cysteamine therapy. Second, we will apply advances in opensource, artificial intelligence-driven computer vision to automatically recognize and quantify cystine crystals with greater accuracy and robustness than existing proprietary methods, ultimately improving the accessibility of this imaging technology. Finally, we will then correlate skin cystine crystal burden with measures of other organ function such as the kidney and thyroid.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

This project will contribute to the further development and increased accessibility of skin reflectance confocal microscopy as a non-invasive method of monitoring cystinosis disease progression and the effectiveness of treatments. This will hopefully enable even more personalized therapeutic adjustments.

ANTICIPATED OUTCOME:

We anticipate defining the natural history of skin cystine crystal accumulation in cystinosis while on systemic cysteamine. Furthermore, we expect to create a robust, open-source, and AI-powered software that accurately quantifies this crystal burden. By eliminating the need for expensive software, this outcome will make the technology more accessible, firmly establishing skin RCM (reflectance confocal microscopy) as an invaluable biomarker and companion diagnostic for guiding cystinosis therapies.

THE IMPACT *of* CRF RESEARCH

SINCE 2003, THE CYSTINOSIS RESEARCH FOUNDATION HAS RAISED AND COMMITTED MORE THAN $73 MILLION FOR CYSTINOSIS RESEARCH, MAKING CRF THE LARGEST PROVIDER OF GRANTS FOR CYSTINOSIS RESEARCH IN THE WORLD. OUR DEDICATED RESEARCHERS AND SCIENTISTS ARE WORKING IN 13 COUNTRIES AROUND THE WORLD TO FIND BETTER TREATMENTS AND A CURE FOR CYSTINOSIS.

TO SEE EACH AREA OF RESEARCH FOCUS & GRANTS AWARDED IN DETAIL, VISIT:

WWW.CYSTINOSISRESEARCH.ORG/IMPACT

AREAS OF RESEARCH FOCUS & GRANTS AWARDED SINCE 2003

Cystine Measurement and Cysteamine Toxicity
10 GRANTS

Thyroid
1 GRANT

Cellular and/or Molecular Studies of the Pathogenesis of Cystinosis
69 GRANTS

Skin, Muscle and Bone
23 GRANTS

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

Molecular Study of Cystinosis in the Yeast Model
3 GRANTS

New Drug Discovery Cysteamine, New Medications and Devices
32 GRANTS

Genetic Analysis of Cystinosis
5 GRANTS

Neurological
17 GRANTS

Cure Cystinosis International Registry (CCIR)
1 GRANT

Eye-Corneal Cystinosis Research
12 GRANTS

Rat Model for Cystinosis
4 GRANTS

Kidney Research
30 GRANTS

Lab Equipment for Cystinosis
10 GRANTS

CYSTINOSIS RESEARCH FOUNDATION

SCIENTIFIC REVIEW BOARD

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

CHAIRPERSON

Stéphanie Cherqui, PhD
Professor, Department of Pediatrics, Division of Genetics
University of California, San Diego
SAN DIEGO, CALIFORNIA

THANK YOU FOR THE YEARS OF DEDICATION TO THE CYSTINOSIS COMMUNITY.

BOARD MEMBERS

Sergio D. Catz, PhD
Professor, Department of Molecular Medicine
Scripps Research Institute
LA JOLLA, CALIFORNIA

Olivier Devuyst, MD, PhD
Professor, Institute of Physiology
Head of Mechanisms of Inherited Kidney Disorders Group
University of Zurich
ZURICH, SWITZERLAND

Francesco Emma, MD
Head of Pediatric Nephrology, Director of Nephrology Laboratory
Bambino Gesù Children's Hospital
ROME, ITALY

Benjamin Freedman, PhD
Assistant Professor, Department of Medicine, Division of Nephrology
University of Washington
SEATTLE, WASHINGTON

Larry Greenbaum, MD, PhD, FAAP
Division Director of Pediatric Nephrology Bernard Marcus Professor of Pediatric Nephrology, Emory School of Medicine
Executive Clinical Director, Children's Healthcare of Atlanta
ATLANTA, GEORGIA

Paul C. Grimm, MD
Professor of Pediatrics, Medical Director
Stanford Children's Health Pediatric Kidney Transplant Program
Stanford University School of Medicine
PALO ALTO, CALIFORNIA

Julie R. Ingelfinger, MD
Deputy Editor of the New England Journal of Medicine
Professor of Pediatrics
Harvard Medical School
BOSTON, MASSACHUSETTS

Stephen L. Jenkins, MD
Assistant Professor, Hospitalist, Department of Internal Medicine
University of Utah Hospital
SALT LAKE CITY, UTAH

Aude Servais, MD, PhD
Senior Nephrologist, Department of Adult Nephrology
Necker Hospital, Paris Descartes University
PARIS, FRANCE

BOARD OF TRUSTEES

CHAIR OF THE BOARD
Nancy J. Stack
Founding Trustee, President
Cystinosis Research Foundation

VICE CHAIR
Geoffrey L. Stack
Founding Partner
SRG Partners

TREASURER
Donald L. Solsby
Compass Direct

BOARD MEMBERS

Marcu Alexander
Founder
Hearts for Hadley

Stéphanie Cherqui, PhD
Professor
University of California, San Diego

Clay Emerson PhD, PE, CFM
Sr. Technical Director
Princeton Hydro, LLC
Hope for Brooke

Jill Emerson, CPA
Hope for Brooke

Denice Flerchinger
Founder
Tina's Hope For A Cure

Stephen L. Jenkins, MD
University of Utah Hospital
Sam & Lars Hope for a Cure

David W. Mossman
Aztec Mustang

Kristen Murray
Seth's Circle of Hope

Kevin Partington
Executive Director
Cushman & Wakefield
Jenna & Patrick's Foundation of Hope

Teresa Partington
Founder
Jenna & Patrick's Foundation of Hope

Brian Sturgis
Senior Director
SimulStat Incorporated
24 Hours for Hank

LEADERSHIP.
GUIDANCE.
COMMITMENT.

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 $73 million with 100% of your donations going to support cystinosis research.

EDUCATION

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

cystinosis RESEARCH FOUNDATION

MEDICAL AND SCIENTIFIC ADVISORY BOARD

Bruce A. Barshop, MD, PhD
University of California, San Diego
SAN DIEGO, CALIFORNIA

Stéphanie Cherqui, PhD
University of California, San Diego
SAN DIEGO, CALIFORNIA

Ranjan Dohil, MD
University of California, San Diego
SAN DIEGO, CALIFORNIA

Laurence Greenbaum, MD, PhD
Emory University School of Medicine
and Children's Healthcare
ATLANTA, GEORGIA

Paul C. Grimm, MD
Lucile Packard Children's Hospital,
Stanford University School of Medicine
PALO ALTO, CALIFORNIA

Stephen L. Jenkins, MD
University of Utah Hospital
SALT LAKE CITY, UTAH

Robert Mak, MD, PhD
University of California, San Diego
SAN DIEGO, CALIFORNIA

Julian Midgley, BM, BCh
Alberta Children's Hospital
CALGARY, AB, CANADA

Doris Trauner, MD
University of California, San Diego
SAN DIEGO, CALIFORNIA

Zoe R. Solsby
Vice President

WE'RE BETTER TOGETHER

cystinosis RESEARCH FOUNDATION

Our collective of donors, researchers, and community partners unite for a future free from cystinosis.

SCAN TO DONATE

HELP ACCELERATE OUR RESEARCH TODAY!