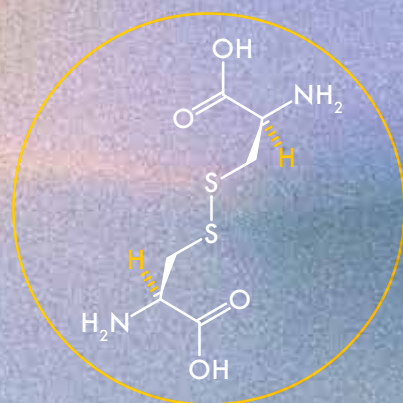




Generation HOPE

CURES
BEGIN
HERE

S U M M E R 2 0 2 6

Generation HOPE!

CURES BEGIN HERE



Cystinosis Research Foundation
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CYSTINOSISRESEARCH.ORG



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Please send suggestions and comments regarding
Cystinosis Magazine to nstack@cystinosisresearch.org

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The mission of the
Cystinosis Research
Foundation is to find
better treatments and
a cure for cystinosis by
supporting bench, clinical,
and translational research.
Since 2003, CRF has
raised \$73 million for
cystinosis research in
an effort to find a cure.

S U M M E R 2 0 2 6

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A LETTER FROM NANCY AND JEFF STACK



Dear Family & Friends,

As summer unfolds, we are filled with a sense of lightness and possibility, and we are reminded of the strength and connection that defines the cystinosis community. At the Cystinosis Research Foundation, this season reminds us not only of how far we have come, but also of the extraordinary community that makes every step forward possible. Because of you: our families, researchers, and supporters, we continue to move closer to better treatments and, a cure for cystinosis.

CRF began with a simple wish from Natalie “to have my disease go away forever.” From that wish, grew a foundation grounded in hope and determination. Over the years we have built an international research community focused on improving the lives of those with cystinosis. What began as one voice has grown into a collective effort to find a cure for cystinosis. CRF has connected families, inspired researchers, and built a community of strength and shared purpose that continues to change lives.

a quiet strength, a deep love, no one in
this community walks the journey alone

CYSTINOSIS MAGAZINE HIGHLIGHTS

In this issue, we are proud to introduce two researchers: Dr. Umber Dube and Dr. Veenita Khare, both of whom work with Dr. Stéphanie Cherqui at UC San Diego. Dr. Cherqui's lab is committed to a comprehensive approach as it seeks to transform the lives of cystinosis patients.

Dr. Dube's study is based on data collected from 90 patients at the Day of Hope family conferences. The data is the largest data set of skin reflectance confocal microscopy in the world. The aim of Dr. Dube's research is to help with diagnosis and cysteamine dosing.

Dr. Khare is developing new mouse models that will be invaluable tools for researchers around the world. Dr. Khare hopes that the new mouse models will mimic the human phenotype more closely, expanding the exploration of novel treatments. The current animal models have shown a "genetic drift" with a particular loss of the kidney phenotype. Creating a mouse that exhibits Fanconi syndrome is the objective of her study.

We have so much to share and to celebrate but we know that the heart of our magazine lies in the stories of our families and patients. Each story is unique, but they all share something important—a quiet strength, deep love, and the reminder that none of us in this community walks this journey alone.



UPDATE ON THE NOVARTIS CYSTEM CLINICAL TRIAL

We are closer than ever to realizing a cure. Novartis has opened the next phase of the CYStem stem cell and gene therapy clinical trial for children ages 2–5 with cystinosis. We are thrilled to report that the first patient was accepted into the trial just before her sixth birthday and was transplanted in mid-May.

This phase will evaluate the safety, tolerability, and effectiveness of this groundbreaking treatment, allowing some of our youngest patients to participate for the first time. Currently, there are three clinical sites open; Emory University (Cohort 0), where patients will continue standard of care to provide essential comparison data, Stanford University, and UC San Diego (Cohort 1), where patients will receive the stem cell and gene therapy treatment.

This progress is rooted in the vision of Dr. Stéphanie Cherqui. In 2007, CRF took a bold step in funding her work, believing in the potential of her research. That decision was an investment that led to the first clinical trial in 2018 and today's expansion into pediatric trials. Her work

has transformed the landscape of cystinosis research. We are deeply grateful to Dr. Cherqui for her leadership, perseverance, and unwavering dedication that continues to inspire and guide us forward.

The current trial would not exist without the five adult patients who participated in the early, first phase of the trial at UC San Diego. We are forever grateful to them — their participation pioneered the stem cell trial for the cystinosis community.

The Novartis trial is a critical and meaningful step forward—one that reflects years of shared commitment, perseverance, and belief. If the Novartis trial proves successful, we anticipate Novartis will seek accelerated approval for the treatment for all people with cystinosis. We now stand on the threshold of a potential therapy that could slow disease progression or even lead to a cure.

A complete list of the inclusion/exclusion criteria for the trial can be found on the CRF website, www.cystinosisresearch.org/current-clinical-trials/ and on clinicaltrials.gov/.

>>>>

CURES BEGIN AT CRF

The progress we are seeing today is built on years of steady investment in research. Since 2003, CRF has funded 255 multi-year research studies in 13 countries, helping to drive discoveries that have led to over 122 published articles in prestigious scientific journals. Our policy of awarding research grants bi-annually, guarantees that there are no gaps in funding life-saving research. We are grateful to the CRF Scientific Review Board, who thoughtfully evaluates every research application we receive and recommends the most promising research studies to fund.

The research you support has led to two FDA approvals: Procybsi® (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.

We are pleased to report that CRF funded three research studies in the fall of 2025, totaling \$621,830. The studies

there are no gaps in
funding life-saving research

explore several important research areas. One study examines the complex causes of bone disease in cystinosis. Another study investigates whether muscle weakness in cystinosis is due to dysfunction in muscle stem cells responsible for regeneration. A third one continues ongoing work, testing whether LNP/mRNA therapy can prevent or even reverse kidney disease in cystinosis mouse models. Additionally, a new area of research explores ferroptosis, an iron-driven form of cell death and whether it contributes to kidney damage. The research abstracts for these studies can be found on pages 45-47.

LOOKING AHEAD

As we enter this next phase of clinical trials, we are reminded that the impact of our research extends far beyond cystinosis. Discoveries supported by CRF have advanced understanding in other diseases, including Friedreich's ataxia, Alzheimer's, and other genetic and systemic diseases like cystinosis. While our focus remains firmly on cystinosis, we are proud that our work is helping to create pathways to cures for other communities as well.

We have so much to be grateful for—your steadfast support, your belief in the power of research, and your compassion and love you show this community. Together, we have built something truly extraordinary: a global community united by hope, dedicated to improving lives and finding a cure for those living with cystinosis.

Our mission continues, and with your partnership, we will keep pushing forward with purpose and determination, driving discovery, advancing critical clinical trials, and bringing renewed hope to every family affected by cystinosis. CRF is where cures begin, and we truly believe that a cure is within reach.

Thank you for being part of the CRF family, and for walking along side Natalie and all those living with cystinosis. Your compassion, commitment, and support mean more to us than we can express, and your belief in our work inspires us every day to continue our mission.

With heartfelt gratitude,

Nancy and Jeff



2026 has already been a year of reflection, gratitude, and hope for our family. This past year brought incredible change—we moved back to Southern California to be closer to family, and shortly after, on July 24, 2025, we welcomed our beautiful daughter, Isabelle Christina Morgan, into the world. Being back in sunny California and reconnecting more regularly with family and friends has brought us so much joy.



Now at nine months old, Isabelle is thriving—full of joy, curiosity, and personality. She loves to say ‘dada’ and go on family walks around our neighborhood with our dog, Wesley. She is 22 pounds and growing like a weed! Becoming her mom has been the greatest gift of my life and has strengthened my determination to stay as healthy as possible so I can be there for every moment ahead.

At 35 years old, I continue to do well, other than my kidney function. It continues to decline, and I am still waiting for a kidney transplant. I see my nephrologist regularly and undergo blood work every three months to monitor things closely. While this season comes with uncertainty, it is also filled with hope. One of the greatest sources of that hope is my husband, Danny, who made the selfless decision to enter the kidney donor match program so that I can receive a kidney through a living donor chain. His love and sacrifice mean more than I can ever fully express and have brought us closer to the reality of a successful transplant. My goal is to avoid dialysis while I wait for the right match.

I continue to be under the care of an incredible transplant team and remain deeply grateful for the support surrounding me. This March, I also celebrated my “fourth birthday” since my stem cell and gene therapy transplant in 2022—a milestone that reminds me how far I’ve come and how much hope there is for the future. I remain on a very low dose of cysteamine and continue with follow-up care at UCSD. The stem cell transplant was life-changing for me, and I hope that one day it will be accessible to everyone living with this disease. I have faith that the next phase of the stem transplant will be successful and continue to bring hope for a cure in our community.

Thank you to each of you who continue to support me and others living with cystinosis. Your encouragement, advocacy, and belief in a better future have not only helped save my life, but have also given hope to so many others living with 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 255 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

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 although each "orphan" disease affects only a small number of patients, studying one often leads to advances in other rare and more common disorders.

CRF GLOBAL OUTREACH - 13 COUNTRIES

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



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

255 Multi-Year Grants
in 13 Countries

255

PUBLISHED

122 Articles in Prestigious
Scientific Journals by
CRF-Funded Researchers

122

RAISED

More Than \$73 Million
for Cystinosis Research

\$73M

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

IN 2025

CRF Awarded

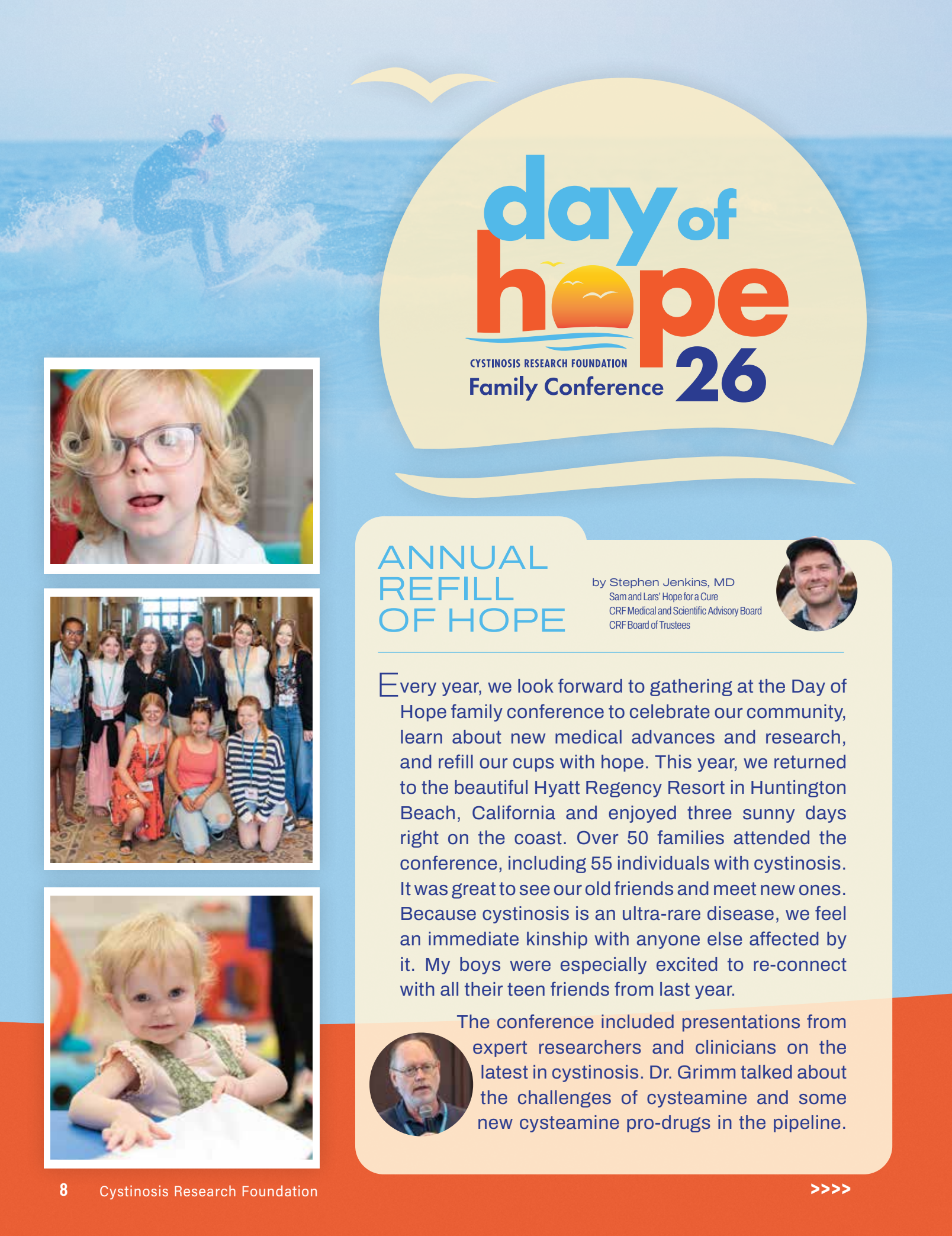
7

New Research Grants

Totaling More Than

\$1,181,024 in 5 Countries

and Published 6 Articles in Prestigious Journals by CRF Researchers



day of hope

26

Family Conference

CYSTINOSIS RESEARCH FOUNDATION

Family Conference

26



ANNUAL REFILL OF HOPE

by Stephen Jenkins, MD
Sam and Lars' Hope for a Cure
CRF Medical and Scientific Advisory Board
CRF Board of Trustees



Every year, we look forward to gathering at the Day of Hope family conference to celebrate our community, learn about new medical advances and research, and refill our cups with hope. This year, we returned to the beautiful Hyatt Regency Resort in Huntington Beach, California and enjoyed three sunny days right on the coast. Over 50 families attended the conference, including 55 individuals with cystinosis. It was great to see our old friends and meet new ones. Because cystinosis is an ultra-rare disease, we feel an immediate kinship with anyone else affected by it. My boys were especially excited to re-connect with all their teen friends from last year.

The conference included presentations from expert researchers and clinicians on the latest in cystinosis. Dr. Grimm talked about the challenges of cysteamine and some new cysteamine pro-drugs in the pipeline.







He also talked about a newer class of medications, “floxins,” that have been found to delay progression of chronic kidney disease. Dr. Olivier Devuyst, a CRF-funded researcher at the University of Zurich, is studying the effects of floxins on Fanconi syndrome in cystinosis. Dr. Grimm also talked about the importance of vaccinations for kidney health and transplant, and advances in transplant medicine.

We heard from a new researcher, Dr. UMBER DUBE, a dermatologist at UC San Diego, about the different ways cystinosis affects the skin and hair. Cystinosis is associated with increased pheomelanin, which raises the risk of skin cancer and skin aging. Using sunscreen can help protect against UV radiation.



Dr. Beno Freedman gave an update on his research of turning urinary cells into stem cells, which he programs into mini-kidney organoids. He injects these organoids into mouse kidneys, where they form more advanced kidney cells and structures. Hopefully one day, this research will lead to gene-edited organoids in humans with cystinosis that could be grafted into native kidneys to improve function.

We heard from a new researcher, Dr. Feodor Price, who is doing similar work with muscle cells. He studies satellite cells, which are a special stem cell that can become skeletal muscle cells. His lab has created muscle organoids, and they are doing extensive analysis on what genes get transcribed and what proteins are synthesized in satellite cells from people with cystinosis, to better understand if there's a way to improve muscle wasting.

Dr. Donny Suh shared an update on his research on the ways cystinosis affects the eyes. He has been doing eye exams for the last three years at the Day of Hope and has collected a lot of data. He has developed a new slit-lamp-based corneal







cystine crystal score that correlates well with more advanced imaging.

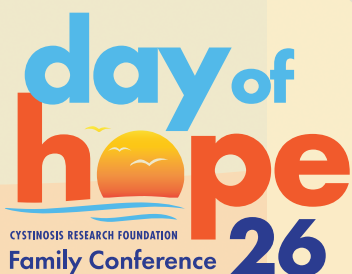
Dr. Reza Sadjadi shared an update on his clinical study of muscle weakness and swallowing problems in cystinosis. He is recruiting patients ages 16 and up to study three different interventions that might improve swallowing and decrease the risk of aspiration. If you are interested, you should contact Dr. Sadjadi!

Dr. Goodyer shared an update on his work developing mRNA treatment for cystinosis. Moderna has created cystinosin protein mRNA transcripts packaged in lipid nanoparticles, which Dr. Goodyer has injected into cystinosis mice with Fanconi syndrome, and after six weeks, the Fanconi syndrome resolved. This could be an incredible treatment option in the future!

The scientific sessions concluded with an update from Dr. Stéphanie Cherqui on the longitudinal data from the adult stem cell trial. Her work was recently published in the most prestigious medical journal in the world, the New England Journal of Medicine. Following Dr. Cherqui, Dr. Wigderson from Novartis gave a presentation explaining the protocol for the CYStem trial which will test gene therapy in children ages 2-5 with cystinosis and Fanconi syndrome.

In addition to scientific updates, we had breakout sessions based on age groups, where parents and people with cystinosis could connect and discuss the challenges and silver linings of cystinosis. We also heard from the Goulet/Provost family about their daughter Zoé, and the Khalasi family about their son, Aarav. It's always inspiring to hear the stories of members in our community and connect based on our shared experiences.

The 2026 Day of Hope was a powerful reminder of how far the cystinosis community has come and how much promise lies ahead. While the challenges of cystinosis remain real, so does the resilience of this community and the dedication of the researchers working toward better treatments and ultimately, a cure. We left Huntington Beach feeling rejuvenated and hopeful. We already miss our friends and can't wait for next year!



THANK YOU FOR HELPING HOPE BLOOM!

Thanks to your relentless support, generosity and hope, we have raised over \$785,726 for cystinosis research! Every donation made, big or small, has helped bring us closer to a world without cystinosis.

What started as just a seedling of an idea, "to have my disease go away forever," has flourished into a global, research-driven effort to create a brighter, more hopeful future for all those affected by cystinosis. With your care, commitment, and cultivation, Natalie Stack's wish for a cure is no longer just a dream, but a possibility that is finally within reach.

We have made progress, but much remains to be discovered. Our relentless pursuit of a cure requires that we continue to fund cystinosis research with the greatest potential. Natalie's wish for a cure is our wish, and there is still so much more work to do to make it happen.

It's never too late to donate and support life-saving research. Make your mark today by visiting the Natalie's Wish page on our website.

WWW.CYSTINOSISRESEARCH.ORG/NATALIES-WISH-2026

Natalie's
Wish
26

THE WISH
THAT IS
CHANGING
LIVES

WITH YOUR SUPPORT,
WE RAISED OVER
\$785,726 FOR
CYSTINOSIS RESEARCH



SCAN TO MAKE
A DONATION.

A BIG THANK YOU!

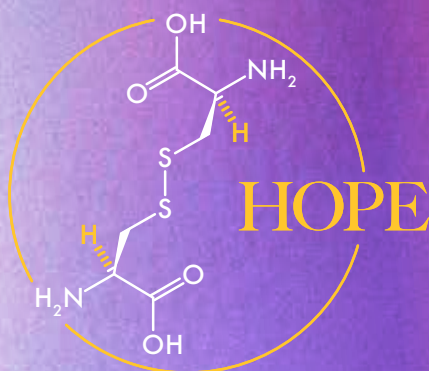
Giving Tuesday Keeps on Giving!

Your generosity this Giving Tuesday was truly extraordinary. Thanks to your unwavering support, we surpassed our matching gift challenge and raised more than **\$284,648** for cystinosis research.

At CRF, 100% of every donation goes directly to advancing research for better treatments and a cure. Because of you, progress is not just possible—it's happening. You are the force behind every breakthrough, every milestone, and every step forward.

Your commitment to the children and adults living with cystinosis fuels our mission to create a world without this disease. Thank you for making this momentum—and this hope—possible.

— 2025 —
**GIVING
TUESDAY**



SUMMER 2026

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GENERATION HOPE!



CYSTINOSIS COMMUNITY CALENDAR OF EVENTS

We would like to acknowledge all families for their support of cystinosis research.

Saturday, July 11, 2026

HOPE FOR JAMES GOLF TOURNAMENT
Valley Regional Park, Rosthern, Saskatchewan, Canada
Email: hopeforjames09@gmail.com

Saturday, October 21, 2026

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

Saturday, November 7, 2026

**6TH ANNUAL COLLINS CURE CUP
GOLF TOURNAMENT
IN HONOR OF COLLINS GALLOWAY**
Windermere Golf Club
For Information: www.collinscure.org/golf-tournament

Tuesday, December 1, 2026

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

February 18 and 19, 2027

**TENTH CRF INTERNATIONAL
CYSTINOSIS RESEARCH SYMPOSIUM**
Pacific Club, Newport Beach
By Invitation Only

Sunday, February 28, 2027

RARE DISEASE DAY
Cystinosis Research Foundation
www.cystinosisresearch.org

Spring 2027

**ANNUAL FISHING FOR BROOKE'S CURE
IN HONOR OF BROOKE EMERSON**
Hope for Brooke
Contact Clay Emerson: cemerson@cystinosisresearch.org

Spring 2027

**ANNUAL PICKLEBALL TOURNAMENT
IN HONOR OF ISLA McALLISTER**
Contact Duncan McAllister: duncanmcal@yahoo.com

Month of April 2027

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

To Be Determined

CRF DAY OF HOPE FAMILY CONFERENCE
Contact Clay Emerson: cemerson@cystinosisresearch.org

WHERE CURES BEGIN

A TRIBUTE TO OUR KIDNEY DONORS

Dear Family and Friends,

The Partington Family is doing well! We have 21-year-old twins at college, each with a newly transplanted kidney. The following are tributes to each of their anonymous donors. We will share the letters with Jenna and Patrick's transplant coordinator, asking her to pass them along. We would love to hear back and learn the story behind these angels out there!

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

At this moment of levity, it's important to point out that a kidney transplant is only a patchwork solution to the problem. A band-aid. A beautiful, life-extending gift, but not enough to keep cystinosis from progressing to the muscles, bones, and other organs of the twins' bodies. We knew the time for kidney transplant would come, just like we know cystinosis is progressive, and sooner or later our children's health will be compromised in new, devastating ways. Our work is not done, and we will continue to support the Cystinosis Research Foundation and its efforts toward improved treatments and a comprehensive cure for cystinosis. We are impressed with the discoveries being made and the carefully executed clinical trials underway. At one time, Jenna and Patrick were too young to participate in early-stage trials, and now they find themselves too old for current trial parameters. Like thirsty souls walking toward a mirage of water: they see it ... they want it ... and they NEED it. We have to trust there is a reason it eludes them.

Read the letters
for the donors on
the next page.



A TRIBUTE TO OUR KIDNEY DONORS

January 29, 2026

Dear Patrick's Kidney Donor,

I am Teresa, mom to twin children, Jenna and Patrick, who are 21. Today I'm at Stanford Medical Center with my husband Kevin, and with Patrick, who is having what we hope will be his final dialysis treatment in a long, long time! Patrick is scheduled to receive a healthy kidney (which you have carried for him all these years) sometime after 3 pm today. We are anxious and eager and grateful and prayerful. What a gift. What a beautiful gesture to be part of.

Patrick and his twin sister have a genetic disease that affects the kidneys very early in life, so we have known for years that this day would eventually come. Two months ago, Patrick had both of his native kidneys removed, as they were quite sickly. He has healed from that surgery and has had dialysis three times a week for four hours each visit. He has handled it valiantly. It would not be surprising to learn that your gift has been given in the name of someone who knows dialysis all too well. I hope your contribution to Patrick, specifically—and the National Kidney Registry—is one that brings complete relief to the person you are giving it for.

Our family story is made more complex by the fact that our daughter Jenna received a kidney through the National Kidney Registry just last August 19. I have not yet written to her donor; I was too anxious to consider typing that day! Kevin donated to her in December of 2024, and after 24 hours in her body, it had to be removed due to renal vein thrombosis. Exceedingly rare, but utterly devastating. Jenna was on dialysis for 18 months while another loved one donated in her honor and earned her a voucher for her second transplant. Jenna is now back at college in Idaho and is THRIVING. Knowing that this is possible for Patrick is thrilling, and you should know what a special part you play in this story!

Patrick loves nature, movies, and time with friends. He loves San Francisco, where he will return to college when he feels better. He is going to take great care of the gift you've so selflessly given!

With so much Gratitude!

Teresa



we would love to hear back and learn
the story behind these angels out there

The Abiding Gift of "Tammy Sparkles"

On August 19, 2025 a well matched kidney arrived to our daughter, Jenna McCall Partington, at Stanford University Medical Center. After a harrowing year of a failed and emergently removed transplant from Kevin, nearly a year of dialysis, and three semesters away from college; the gift came from an unknown angel who donated on behalf of their loved one via the National Kidney Registry. It was packed on ice into a cooler, escorted away from its donor on the East Coast via a commercial flight, then by car along California's beautiful I-280 freeway, from airport to operating room.

The making of urine is the sign of success for kidney transplant, and within 30 seconds of Jenna's renal vein being connected to her new kidney, she was making urine. After my joyful visit with Jenna's surgeon that day, it was fun to text everyone waiting to hear the news of Jenna's operation with this image:

Naming a new organ and celebrating its birthday is common in transplant circles, and Jenna's kidney is called "Tammy Sparkles." A rather random title bestowed during a phone conversation Jenna had with her cousin Lindsey soon after transplant.



In elementary school Jenna and Patrick had a "Flat Stanley" project, based on the children's book by Jeff Brown, which showed a figure made of construction paper (named Stanley) traveling around the globe, exploring new places, and having new adventures. I can't help but think of Tammy Sparkles as a human organ version of Stanley, with many new travels to be celebrated and documented. We will pass this CRF magazine submission along to Jenna's donor just as we would a Flat Stanley project, sharing recent adventures.

Tammy Sparkles resides in the little college town of Moscow, Idaho, in a single bed/bath apartment that has a baby pink corduroy sofa, F1 posters, and green vintage kitchen dishware, purchased at thrift stores. She has blessed her new owner, Jenna, with the health and energy to learn how to cook new foods, such as her favorite: Crispy Brown Butter Gnocchi with Brussels Sprouts (NYT Cooking, Ali Slagle). She also entertains friends. But most impressively, Jenna now manages her life-saving medications entirely on her own for the first time, from picking them up at the pharmacy to organizing her pillbox and taking them on schedule.

Tammy Sparkles also fuels Jenna's walks to her classes at the University of Idaho and helps her stay on top of her to-do lists and homework more easily and successfully than ever before. Everyday tasks involving stamina and executive function that were historically insurmountable for Jenna are completed with ease.

Spring break 2026 was spent in Nashville, Tennessee with her dear friend, Natalya. With the energy to travel a long distance, manage airports and schedules independently, and pack and take the proper medications while away, Jenna and her new organ enjoyed the food, music, and a social scene that were a dream one year ago when dialysis was just about all Jenna could muster the strength for.

This summer, Tammy Sparkles will enjoy her first visit to McCall, Idaho, the town that is Jenna's middle name and take an inaugural swim in Payette Lake. She will have a full-time job at the University Bookstore ... once again offering the strength and stamina for what was once impossible. She loves independent living and learning and thinking about the future.

Tammy Sparkles is thriving in her new environment. And her host is a remarkable young lady, who cares for her with reverence and gratitude. There is no doubt that these two are going to have a fine time together!

Building a Family When Your Child Has Cystinosis

When a child is diagnosed with cystinosis, life changes in a way parents could never have imagined. The diagnosis brings a wave of emotions: fear, uncertainty, grief, but also determination and deep love. Families quickly learn how to manage medications, appointments, and the many daily routines needed to keep their child healthy.

Amid these new responsibilities, another deeply personal question may arise over time: Can we still grow our family?

For us — and probably for many parents of children with cystinosis — the idea of having more children brings both hope and uncertainty. After the initial diagnosis, we put our future plans on hold while adapting to our new reality. As time passes and routines become more familiar, we've begun to feel ready to think about the future again. Concerns about genetics risks, time, energy, and emotional capacity are very real. Yet many families do find ways to continue building their families, each in their own way. Why couldn't we then? There is no single right answer — only the path that feels right for each family. Families have several possible paths forward, and each comes with its own considerations. These reflections are not obstacles — they are part of thoughtful parenting.

No family should face these decisions alone. Support can come from many places: medical teams, genetic counsellors, extended family, and patient organizations such as the Cystinosis Research Foundation.

Connecting with other parents in the cystinosis community can be especially reassuring. Hearing how others have navigated similar questions reminds families that there is no perfect decision — only the one that feels right for them.

For our own family, it became a long and emotional journey. After Juliette's diagnosis, we spent five years pursuing IVF with genetic testing, hoping to avoid the genetic lottery and start a pregnancy with a healthy embryo. We had multiple unsuccessful attempts, and the

process required patience, resilience, and many moments of doubt and disappointment.

Eventually, what we tried to avoid for years happened: we became pregnant naturally.

What followed was another intense period of waiting. Through prenatal testing, we had to wait to learn whether the baby would be affected. Those weeks felt incredibly long, filled with hope but also fear.

When the results finally came, they brought immense relief: the baby had taken the best of our genes! After years of uncertainty and effort, it truly felt like a big miracle.

Our story is only one among many, but it reminded us that family journeys rarely follow a straight path, and that there is always hope for better days.

In our family, although adjusting to this new "structure" of four was not always easy, we quickly realized that siblings could become a powerful source of connection and strength. Juliette had longed for a little sister, and she has since developed a remarkable sense of responsibility, along with deep love and a natural nurturing instinct. We are convinced that our daughters will grow up supporting each other and this will be a lasting source of strength for them.

What we shall always keep in mind is that cystinosis may shape family life, but it does not define it. Families touched by cystinosis often discover strengths they never expected — resilience, compassion, and a profound appreciation for each moment together.

Whether a family chooses to have more children, adopt, or focus on the child they already have, the most important truth remains the same: A family is not defined by numbers, but by the love, care, and support shared within it.

And within the cystinosis community, that love is remarkably strong.



we've begun to think
about the future again



the stem cell transplant

By Kurt Gillenberg, 33 years old
LEANDER, TEXAS



It's hard to believe it's been almost four years since my stem cell transplant. Time has flown by, and yet so much has happened in that time. I've changed jobs twice, had open heart surgery, moved halfway across the country—and to top off 2025, I experienced a DKA episode in early December.

four years later

I'm currently back home in Texas after traveling to San Diego for my stem cell follow-up. These visits always take some mental preparation. There's still testing involved, and since I no longer live in California, the travel can be a bit of a process. But honestly, I don't mind. It gives me a chance to get out of the house for a few days, reset a bit, and most importantly, spend time with my grandpa while I'm there. Over the last few years, these follow-ups have become much easier. There's less testing now, and instead of being there for several days, everything can usually be done in about two. I'll go back again in six months, and after that, it will move to once a year for the next 13 years.

This past year felt like it flew by while somehow dragging at the same time. For a lot of people I know, 2025 was a difficult year. For me, things were calm . . . until they weren't. The week before Thanksgiving, I left my job to pursue a new opportunity. It felt like a fresh start and the right decision at the time. But about a week and a half later, everything changed.

I went into DKA—Diabetic Ketoacidosis—which is something I had never experienced before. One night in early December, I went to bed feeling completely normal. During the night, I woke up several times, thinking I was dreaming, when in reality I was hallucinating. The next morning, my parents found me unresponsive and immediately called 911. I was rushed to the hospital with a blood sugar level over 500. My family was told I was extremely lucky I hadn't gone into a diabetic coma.

Doctors believe a bacterial infection likely triggered the episode, but it also revealed underlying diabetic issues that I hadn't been fully aware of. I spent four days in the

hospital getting my blood sugar stabilized and clearing the infection. It was a scary experience—not just for me, but for my family as well—and definitely a wake-up call.

Since then, I've been on a new routine that includes nightly insulin and a much healthier diet. I've cut out

most fast food and junk food, and the difference has been noticeable. I have more energy, I feel better overall, and my workouts have improved. Honestly, compared to everything else I've had to do to stay healthy over the years, this feels manageable.

Looking back now, I realize that the way I had been feeling for a couple of months before the DKA episode was probably my blood sugar being out of balance. It's one of those things you don't fully recognize until something forces you to stop and pay attention. As difficult as the experience was, it pushed me to take better control of my health.

Now that I feel like I'm back in a good place, it's time to focus on what's next. After a lot of thought and many conversations with my parents, I've decided to go back to school to pursue a degree in Radiology and become a radiology tech. I'm 33, and I know I still have the time to build something meaningful for my future. I'm incredibly grateful to have the support system that allows me to take this step. Right now, I'm looking for a part-time job so I can balance work and school when I start, hopefully this summer.

Living with cystinosis has taught me that sometimes life throws things at you that are completely out of your control. It can force you to pause, reset, and focus on healing before moving forward again. That doesn't mean you're falling behind—it just means you're taking the time you need.

There's nothing wrong with picking yourself up, adjusting your path, and getting back out there—no matter how many times you have to do it.

it's time to focus on what's next

Cillian's Journey

C O N T I N U E S ...

By Stacey Maw, Cillian's mom
CAMBRIDGE, ONTARIO, CANADA

Cillian is still absolutely flying!

Cillian's personality radiates off him as he enters a room, always eager to make someone laugh with a joke or a trick. He is such a clever boy, now in GRADE ONE, he has made the adjustment from kindergarten "play" to a structured day at school while remaining a social butterfly.

When Cillian was first diagnosed, I wasn't sure he would even be able to go to school, and now, as we walk up the lane, I hear other little voices shouting his name as he enters the school yard. It's a wonderful feeling, knowing he is thriving at school as kids his age should.

Today, I know we are lucky but, looking back to when Cillian was diagnosed, I never imagined myself saying that, but I know we truly are. All the medications, the strict schedule, the massive amounts of information and all the changes we have made in life have become second nature — our "normal," and our life goes on.

I find myself forgetting most days how much Cillian goes through and how he adjusts so effortlessly. Cillian has always been fiercely independent and, now at seven years old, he is "plugging" himself in, administering his own meds, swallowing pills, and constantly filling his own water bottle, often with a smile on his face. Life would be so much harder if he didn't have such an amazing attitude.

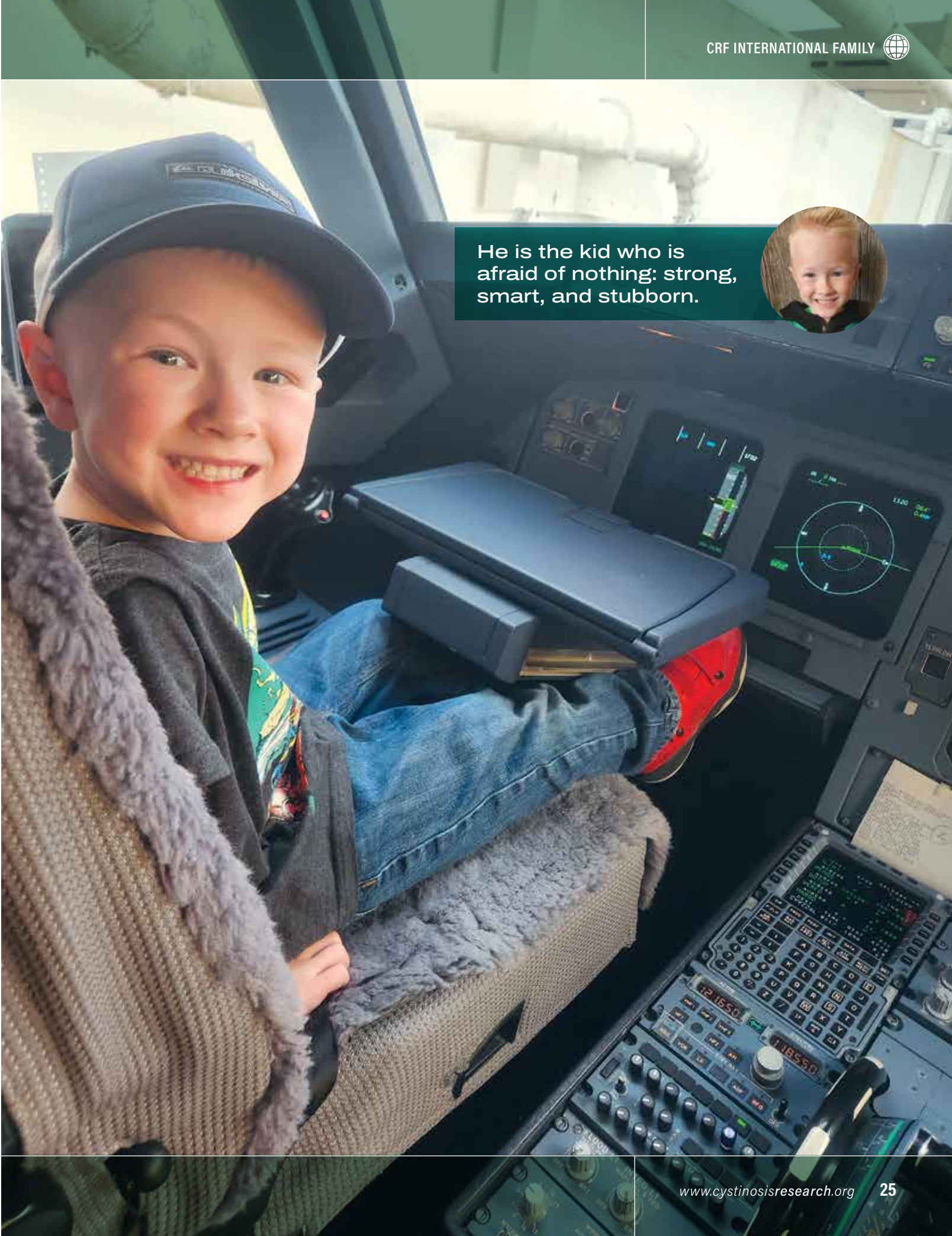
There are days that are still hard. There are times when the common cold has us glued to the couch, away from school and friends. There are days I often feel bad about how fast he has had to grow up and how responsible he has had to become, but I am so beyond proud of him. He has no fear, and nothing holds him back, whether it is ripping around on his 4-wheeler (thanks Pop!) or watching horror movies in the basement with his dad. When the movies make him jump, he laughs; when he falls, he gets up and dusts himself off, unbothered.

Cillian actively pushes through every week with a schedule of school, soccer, karate, and he still finds time for as many crafts as possible. He loves to ride his bike and play outside, whether it is in the back yard or exploring a new park, but also loves a good snuggle on the couch. He is the kid who is afraid of nothing: strong, smart, and stubborn. He has a big imagination, lots of determination and a huge heart; I have no doubt he will do great things.

We are unbelievably grateful to CRF and our cystinosis community for their support and research that allows us to look ahead with hope for what will be Cillian's bright future.



He is the kid who is
afraid of nothing: strong,
smart, and stubborn.



Don't Ever Let Your Disease Stop You

By Samuel Jenkins, 16 years old
MILLCREEK, UTAH

What's up guys?! It's been a while! I'm a junior in high school now. I just got my driver's license, and I took the ACT this year. I take my meds by myself (with the occasional loud reminder from my dad) every day at 7:00 AM, 8:00 AM, 4:00 PM, 7:00 PM, and 9:00 PM. I think it's 62 pills now, but it feels easier when you split it up into 5 doses. Lars, my thirteen-year-old brother, takes his Procysbi® three times a day, which he recommends and says it reduces nausea.

In June 2025, I got knee surgery for my knock knees, a common problem with cystinosis. We were really worried about it going in, but I went to the hospital around 11:00 AM and was out by 3:00 PM. The surgery only lasted an hour, and I felt great afterward. I did need a wheelchair to reach the car, but I was walking distances of 30 feet and even climbing stairs the next morning. The surgery has worked really well! My legs are straighter, and I'm getting the growth implants removed in April 2026.

One of the best things that happened in the last year was that Henry Sturgis moved to Logan, Utah, to major in aviation at Utah State University. Every so often, he stays at our house, and we hang out for a few days. We went camping in southern Utah and went to Lagoon, Utah's only theme park. We've also been skiing with him, which is a blast, and we've gotten into the habit of watching movies till 1:00 AM. He introduced us to *50 First Dates*, and we showed him *Monty Python and the Holy Grail*, a comedic knight movie. I loved *50 First Dates*, but I'm not sure how he felt about *Monty Python*.

I've never been a huge sports person, but when I started school this year, I was bored, so I joined my high school swim team because that's what my friends did. My parents were worried about it, as I had numerous cramps the first week. But we asked my nephrologist about it, and he said it was a good idea and was a great way of staying healthy. I





loved it so much that I decided if I ever got the opportunity to do "Make a Wish," my wish would be to meet and race Michael Phelps. I was pretty bad when I started, but by the end, I had dropped 16 seconds in my 50-yard freestyle, 28 seconds in my 100-yard freestyle, and 13 seconds in my 100-yard backstroke, which is my favorite event. I also swam the 200-yard freestyle. It felt like being hit by a truck while I was swimming it, but the sense of accomplishment outweighed how tired I felt. Needless to say, I was almost as sore as Jake Paul after his fight with Anthony Joshua the next morning.

Another thing I started doing to increase my fitness level was take a weight training class at school. I have it two to three days a week, and I've made lots of new friends who enjoy pumping iron just as much as I do. While I was almost as bad as I was at swimming when I started, (my max deadlift was 95 pounds), I've improved a ton, and now I can deadlift 155 pounds and bench 90. I liked weight training so much that while I had originally signed up for one semester, I decided to take another, and now I'm taking it for my entire senior year as well.

keep trying new things,
even if you don't think
you'll be good at them

I have so much fun at Day of Hope, our annual conference, and I've made boatloads of friends. I couldn't name all of them, but here are some shout-outs to Brooke Carriere, Henry Sturgis, Tina Flerchinger, Seth and Leif deBruyn, Luis Fabian, Karolis Schröder, Hadley and Stella Alexander, and Christian Miller (even though he hit me really hard with a tortilla). At the 2025 conference, I decided to make a documentary to help me remember everything

that happens at Day of Hope. My decision also may have been influenced by my obsession with *The Office*.

It's been a wild few years, and regardless of that one

Taylor Swift song my mom and five-year-old sister are always singing, I am growing up. College is approaching, and I'll have to decide whether to go to Utah State with Henry Sturgis or go somewhere else. But let's be honest: Utah State looks pretty fun. Anyway, my best advice would be to keep trying new things, even if you don't think you'll be good at them. Don't ever let your disease stop you.



Magnolia's story begins in a way that feels familiar to so many families. She was born full-term, and everything appeared exactly as it should — a healthy, happy baby. As the youngest of our five children, Magnolia was a surprise we never saw coming, but from the very beginning, we knew she was a special blessing.

Once we brought her home, though, things didn't feel quite right. Magnolia was often difficult to console and seemed to be sick much of the time. What began as simple concern quickly turned into a long stretch of uncertainty. Before we knew it, we were seeing specialist after specialist, attending countless appointments, and searching for answers that never seemed to come. We found ourselves going down endless rabbit holes, feeling overwhelmed and, at times, defeated.

Everything changed when Magnolia became sick with her first cold. What initially seemed like a routine illness led to bloodwork that revealed her electrolytes were severely imbalanced. That moment set off a whirlwind. Within just 30 days, with two different hospitals working around the clock, we finally received a diagnosis in February 2024: cystinosis.

life-changing

By Kendra Nodland, Magnolia's mom EATONVILLE, WASHINGTON

It was not a diagnosis we could have ever imagined — or one we would have chosen for our daughter. And yet, in the midst of the shock and fear, there was also an unexpected sense of peace. For the first time, we understood what was wrong. We had answers, and more importantly, we had a path forward to help Magnolia.

Since that day, so much has changed for our family. The journey has not been easy — cystinosis comes with daily challenges, unknowns, and emotional highs and lows. But through it all, we have witnessed something remarkable. Our family has grown closer and stronger than ever before. Our faith has deepened in ways we never could have anticipated, anchoring us through every step of this journey.

We have also become part of a community we never knew existed.

The love, support, and shared understanding we've experienced from other families walking similar paths has been life-changing. There is something incredibly powerful about not feeling alone, about learning from others, and about walking alongside people who truly understand.

through it all,
we have witnessed
something remarkable

And in the middle of it all is Magnolia — thriving, growing, and showing us, daily what resilience looks like. Despite the challenges of her diagnosis, she continues to bring joy and light into our home. Watching her reminds us that there is hope — not only for her future, but for so many others living with cystinosis.

One of the most unexpected blessings in this journey has been watching our other children. All of them have embraced Magnolia's story with open hearts. They are eager to share, to educate, and to answer questions from anyone who wants to learn more. Seeing their compassion and understanding grow has been one of the most beautiful parts of this experience.

Cystinosis is not an easy disease, and it has undoubtedly changed the course of our lives. But it has also shaped us in ways we are deeply grateful for. It has strengthened our faith, deepened our relationships, and opened our eyes to a community filled with courage and hope.

What once felt like the end of the unknown became the beginning of a story filled with strength, faith, and unwavering hope.



Our Journey with Our Sunshine Girl!

By Karalee and Nick Zimmerman, Stella Sol's parents
RIVER FALLS, WISCONSIN

Stella Sol was born on February 26th, 2023. It was a magical morning as she was welcomed with the rising sun peeking through our living room windows. Her arriving with the sun would become very fitting because the sun and light always find Stella, even when it feels dark. We even found out we were pregnant on the Summer Solstice.

The first nine months, Stella Sol was a vibrant shining star in our lives. She had an affinity for milk and was the chunkiest baby. She was far from the typical "failure to thrive," meeting every milestone. During Thanksgiving in 2023, when she became ill around nine months old, with little to no improvement for weeks, we decided to take her to the doctor. For the next 40 days, we lived the life that oh so many cystinosis parents have lived: hospital stays coupled with fear and having more questions than answers. After our hospital stay, we left with a g-tube, a laundry list of prescriptions, and you guessed it: a cystinosis diagnosis. We received Stella's diagnosis on December 21, the first day of winter and the darkest day of the year, both literally and figuratively.

Upon diagnosis, we received little to no reassurances from our first care team that she would be okay. Our hearts were shattered, wondering how many birthdays and how many Christmases we would have with her. These were dark days. We felt lost and questioned everything in our lives.

Fortunately, we quickly found answers and hope through CRF and the Day of Hope, along with Stella's magnificently bright light that radiates from her. Although our journey since diagnosis has been riddled with ups and downs, Stella has handled every step flawlessly and her resilience inspires us every minute of the day.

In July of 2024, we unexpectedly became pregnant. Fear and guilt immediately crept in. Although this was a blessing, we were extremely worried. God answered prayers when we did not know where to turn. At 11 weeks pregnant, we underwent intrauterine genetic testing to find out if the baby would have cystinosis. We were ready and prepared for whatever the results were, but were desperately hoping for negative results. At the end of September, we received the best news: that baby did not have cystinosis!

Stella oozes with personality, love, and of course, light.



On March 26, 2025, Graham Wilder Zimmerman entered our world on a nice Spring evening. His middle name comes from the wild journey he had to get here, from a very unexpected pregnancy to the genetic testing—it truly was wild. Graham has been such a blessing to our family and especially to Stella, and he is always nearby at every appointment supporting her.

Although the past year and a half has been filled with ups and downs and life-changing news, we have become well-acquainted to our “new normal.” This new normal is full of appointments, labs, and medications (we call them supplements so Stella knows she is strong and healthy). Stella eats tremendously well, is growing nicely, and hardly

ever throws up (knock on wood!). Stella may have cystinosis, but it does not define her life—or ours. It may impact our lives every second, but it’s simply part of the story.

Stella is truly our shining star. She oozes with personality, love, and of course, light, even when faced with constant poking, prodding, and challenges only those with cystinosis know. Sometimes cystinosis stops us in our tracks. We pause and reflect on the extensive care it requires. But CRF has given us hope — hope for the future and hope for a cure. We strongly believe that the work Nancy, Jeff, and the entire CRF team have done will ensure cystinosis is just a snapshot in Stella’s life and not her entire future.



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!

TOGETHER, WE ARE One

1 PURPOSE.
1 JOURNEY.
1 CURE.

The Emerson Family – Jill, Clay, and Brooke – Hammonton, New Jersey

BROOKE'S 2026 FISHING FOR A CURE

Despite below-freezing temperatures, overnight snow filling the boat, and the challenges of a winter morning, Hans, Ian, Mike, Ralph, and Clay launched with hearts full of purpose: to fight for a cure for cystinosis.



They didn't fish for themselves that day—they fished for Brooke, for every child and family facing cystinosis, and for the hope of a brighter future. Their determination paid off: by day's end, 427 fish had been caught and released, raising more than \$70,000 for research toward better treatments and, ultimately, a cure.

This extraordinary success was only possible because of the love and generosity of the friends and families who stood behind all five fishermen. Every donation, every word of encouragement, and every act of support helped make a real difference.

Brooke, Jill, and Clay are deeply grateful to each fisherman and every donor who believed in their mission. Because of you, hope is stronger, progress is closer, and the dream of a cure is more possible than ever for everyone with cystinosis.



Cystinosis Research Foundation – Irvine, California

RARE DISEASE DAY 2026 - THANK YOU!

Your generosity is the driving force behind every step forward in cystinosis research, and we are deeply grateful for your unwavering support. Because of you, hope continues to grow for children and adults living with cystinosis and for families who dream of a brighter future.

Every gift you make fuels progress, advances groundbreaking research, and brings us closer to better treatments and ultimately, a cure for all those with cystinosis. We are profoundly thankful for your compassion, partnership, and belief in what we can achieve together.

We are proud to continue giving a voice to the rare cystinosis community on Rare Disease Day 2026. Thank you for demonstrating your unwavering commitment to our small but mighty community through your heartfelt donations. Together, we raised more than \$15,000, and every dollar will go directly funding critical global research studies. With your help, we are able to celebrate meaningful progress and renewed hope. Yet there is still important work ahead. While we believe stem cell therapy has the potential to become a life-changing treatment for all people with cystinosis, we must continue investing in research to develop even better therapies for children and adults living with this disease. Our mission is not complete, and your support keeps us moving forward.

Thank you for standing with us on Rare Disease Day and throughout the year. Your generosity is making a real and lasting difference.



TOGETHER, WE ARE One

1 PURPOSE.
1 JOURNEY.
1 CURE.



The Lanken Bonneau Family – Vanessa, Sylvan, and Fera –
Montreal, Québec, Canada

CRI DE NOËL HOLIDAY FUNDRAISER FOR FERA

On the evening of December 22nd, 2025, *Cri de Noël: A Family Christmas Show with the Wainwrights and McGarrigles* raised money for cystinosis research in honour of Fera – a beacon of the family and a little star who loves the stage! Family and friends gathered in Montreal and shared seasonal songs with a festive crowd at la Maison symphonique.

Sylvan and Vanessa wish to thank Martha, Rufus, and the rest of the family, as well as the many guest singers, musicians, and volunteers who gave their time and talent. Together, they helped raise \$35,000 for the Canadian Cystinosis Research Foundation through the Aqueduct Foundation.

Cystinosis families from Canada are committed CRF partners who are dedicated to funding research that will lead to better treatments and a cure for all our children and adults with cystinosis. Thank you!

Fera, stage right, with cousin Martha. Photo by Yagub Allahverdiyev.

The Hughes Family – Emily, Daniel, Adalyn, Carson – Yulee, Florida

A CURE FOR CARSON INAUGURAL GOLF TOURNAMENT

Carson was a healthy, thriving baby who met every milestone until just before his first birthday, when his family's world suddenly changed. After a hospitalization and many unanswered questions, Carson was diagnosed with Fanconi Syndrome and then cystinosis. While the diagnosis was heartbreaking, the Hughes family has chosen hope over fear—drawing strength from the remarkable progress in research and the promise of new therapies that bring us closer to a cure. Their journey has deepened their gratitude for each day and inspired them to turn their love for Carson into action. Today, they are passionate advocates for raising awareness and advancing research for everyone living with cystinosis.

On April 11, Emily and Daniel Hughes proudly hosted their first golf tournament, A Cure for Carson, in Kingsland, Georgia, benefiting the Cystinosis Research Foundation. The inspiring event united 72 enthusiastic golfers and nearly 50 additional guests, all coming together in honor of Carson and everyone living with cystinosis. The day began with breakfast and a shotgun start, followed by 18 holes of golf, a luncheon, exciting raffle prizes, and family-friendly activities for children. The first-place foursome received \$1,000, while GPS rangefinders were awarded for longest drive and closest to the pin.

Thanks to the extraordinary generosity of their community, sponsors, family, and friends, the inaugural event raised an incredible \$28,645 for cystinosis research. Every dollar brings us closer to better treatments and, ultimately, a cure. We are deeply grateful to the Hughes family and everyone who supported this special day. Your compassion, commitment, and belief in a brighter future are helping change lives. Thank you!



TOGETHER, WE ARE One

1 PURPOSE.
1 JOURNEY.
1 CURE.

The Maher Family – Amelia, Tim, Ayla, and Otto – Roslindale, Massachusetts

ALL OUT FOR AYLA AND OTTO

On Saturday, March 28, the Maher family once again brought their community together for “All Out for Ayla and Otto” at Roundhead Brewery in Hyde Park, MA—a day filled with connection, joy, and purpose. Friends, family, teachers, doctors, and coworkers came from every corner of their lives, in a powerful show of support for Ayla and Otto. Their school community turned out in full force, with classmates from first and fourth grade, families, beloved teachers, and even the school nurse joining the celebration.

Laughter and energy filled the venue as guests enjoyed delicious pizzas, empanadas, and refreshments, while children—and plenty of adults—delighted in face painting and arcade games. Beneath the fun was something even more meaningful: a shared commitment to raising critical funds for life-changing cystinosis research.

By the end of the day, Amelia and Tim were left with full hearts and deep gratitude, uplifted by the incredible community standing beside them and inspired by the hope that lies ahead. Thanks to their dedication and the generosity of everyone involved, they raised more than \$48,380 in honor of Ayla and Otto.

We extend our heartfelt thanks to the Maher family for their time and commitment and to their remarkable community for showing up and sharing their love and support. Together, we are unlocking the possibilities of a future without cystinosis!



The McAllister Family – Ashley, Duncan, Graham, and Isla – Edmond, Oklahoma

3RD ANNUAL PICKLEBALL TOURNAMENT IN HONOR OF ISLA

In December 2025, the McAllister family hosted their third annual pickleball tournament benefiting CRF in honor of their daughter, Isla, and all those living with cystinosis. Held once again at Life Time Athletics in Oklahoma City, the event was a joyful celebration of family, community, and hope. Loved ones traveled from Scotland and across the United States to take part, making this year's tournament especially meaningful.

The courts were filled with energy, laughter, and friendly competition as players rallied together for a cause bigger than the game itself. Additional courts were added so children could join the fun—an inspiring reminder that the next generation is already stepping forward to make a difference, and perhaps the beginning of a future children's tournament tradition.

We are deeply grateful to the McAllister family for their unwavering commitment and generosity. Since launching their inaugural pickleball fundraiser in 2023, they have raised more than \$28,495 for cystinosis research in honor of Isla. Every dollar brings us closer to better treatments and, ultimately, a cure.

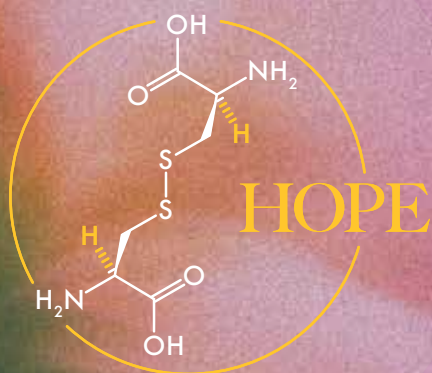
Because of families like the McAllisters, hope continues to grow, progress continues to accelerate, and the future looks brighter for those with cystinosis. Thank you!



SUMMER 2026

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The NEW ENGLAND JOURNAL of MEDICINE

We are honored to share extraordinary news with our community:
Dr. Stéphanie Cherqui's groundbreaking paper,

“HEMATOPOIETIC STEM-CELL GENE THERAPY FOR CYSTINOSIS”

has been published in the New England Journal of Medicine (NEJM).

The NEJM is one of the most prestigious and influential medical journals in the world. With more than 16,000 submissions each year and an acceptance rate of approximately 5%, publication in the NEJM is widely regarded as a hallmark of academic success in medicine. The journal publishes high-impact clinical research and is read by more than a million people globally. Just imagine – the cystinosis story is now being shared around the world!

The CRF story began in 2003 with small, dedicated group of parents who wanted to fund research to improve the lives of their children. At the time, only a handful of researchers were studying cystinosis. In 2006, we met a brilliant scientist, Dr. Stéphanie Cherqui, who believed gene therapy might one day cure cystinosis. We were in uncharted territory but we believed in Stéphanie's vision. Our families and friends believed too and joined the quest. It was, in many ways, the perfect alignment of determination and courage: relentless parents, a committed researcher, brave patients, and a foundation willing to take bold risks in pursuit of a cure.

The publication of this paper represents a significant personal and professional milestone for Dr. Cherqui, but it is also far more than academic recognition. It marks a significant scientific milestone and reflects the extraordinary persistence and excellence of Dr. Cherqui and her colleagues. For the CRF community, it is validation of more than two decades of unwavering commitment to research.

To us, Stéphanie is not only an exceptional scientist, she is a friend. Her work has given our families an immeasurable gift: hope. Hope that we can stop the progression of cystinosis. Hope that this therapy may one day be a cure.

As Novartis oversees the next phase of the trial, we are filled with anticipation as the first patient begins the treatment and remain deeply committed to ensuring that this therapy becomes available to every person with cystinosis.

We congratulate Dr. Cherqui and her team, and we extend heartfelt gratitude to every patient, family, and donor who made this achievement possible.

We will keep the momentum moving forward and continue to fund life-changing research that will give hope to those living with cystinosis.



www.cystinosisresearch.org/dr-stephanie-cherqui-published-article-in-the-new-england-journal-of-medicine

A NEW ORGAN OF UNDERSTANDING

by Dennis Arp

BY FOCUSING ON CYSTINOSIS AND THE SKIN, DR. UMBER DUBE OPENS DOORS TO BETTER DIAGNOSIS, TREATMENT AND OUTCOMES.



It's a universal truth of cystinosis that it attacks every organ of the body. Yet, for all the insights generated by the cystinosis research community, we remain largely in the dark about how the disease affects the largest human organ – the one that enwraps us all.

Our skin.

Dermatologist and research professor Dr. UMBER DUBE is eager to add new layers of cutaneous understanding. Propelled by a grant from the Cystinosis Research Foundation (CRF), Dr. DUBE is exploring the natural history of how cystine crystals accumulate in the skin as he also pursues next-level diagnosis and understanding of cystinosis. Among other innovations, he and colleagues are on a path to apply open-source software that will make cutting-edge skin research technology more useful and accessible.

"Skin is our most visible organ, but given the morbidity and mortality from the other organs impacted by cystinosis, historically skin has been put on the back burner of research into the disease," says Dr. DUBE, Clinical Assistant Professor of Dermatology at UC San Diego.

"Because of all the resources the cystinosis community has brought together – the fund-raising, the amazing advances and the sharing of information – we can now think more about ways to thrive and not just survive," he says. "So, as we consider the long-term health of individuals with cystinosis, the skin becomes an important part. We can use the skin as a big, accessible window to try to understand this disease that impacts the whole body."

Dr. DUBE's dermatological explorations of cystinosis are just one more way that the UC San Diego lab of Dr. Stéphanie Cherqui is committed to a comprehensive approach as it seeks to transform the lives of cystinosis patients.

Dr. DUBE's background includes a focus on the skin care of patients with pigmentary disorders and solid organ transplants, so he's a good fit for cystinosis research. Dermatology also has become an important component of Dr. Cherqui's cystinosis research, including during her lab's breakthrough clinical trials advancing stem cell gene therapy.

GETTING A MICROSCOPIC LOOK AT CRYSTALS IN THE SKIN

Dr. Cherqui saw potential in a skin imaging technology called reflectance confocal microscopy (RCM). That's why she and her lab colleagues asked Dr. DUBE to help advance this non-invasive technique, starting when he was a Chief Resident Physician at UC San Diego.

"It's a bit unconventional for a resident physician to take on this kind of research, but when you've got a good justification and a great team in place, you get excited and you go to work," he says.

Confocal microscopy was first developed in the 1950s and has largely been used for "virtual biopsies" of all kinds of skin conditions but especially malignancies such as melanomas. The technology uses a near-infrared laser (830-nm wavelength). The light can penetrate the skin to illuminate tissue and return dozens of images from the surface level on down. Imagery is captured via a handheld device about every 2.6 micrometers along the way.

Those images can be evaluated in real time as well as archived for further study as part of longitudinal research like Dr. DUBE is conducting. One goal of his project is to further develop and refine software that allows for the sophisticated sorting, classifying, and evaluating of images.

For Dr. DUBE, there's a special joy in sharing what the device shows.

"I like to give those we're imaging an immediate chance to see the crystals in their skin," he says. "They feel the disease, so it's good to let them also see it. As a physician, I like using the technology to help people learn and to deepen our connection."



CYSTINOSIS RESEARCH STAR



Umber Dube, MD, PhD

Dermatologist, Assistant Professor of Dermatology

UNIVERSITY OF CALIFORNIA, SAN DIEGO

USING IMAGERY AS A BIOMARKER FOR CYSTINOSIS

Further advancing the technology, the Cherqui lab has developed reflectance confocal microscopy as a biomarker for cystinosis and has demonstrated that the findings correlate with thyroid and kidney function. The lab has collected skin RCM data during each annual Day of Hope conference since 2017, and it also images participants in the stem cell transplant clinical study.

"We now have 243 skin RCM image sets from 90 individuals with cystinosis, which gives us an incredible resource – the largest skin RCM dataset for cystinosis in the world," Dr. Dube says.

With that data and more to come, the Cherqui lab is helping to determine the natural history of how cystine crystals accumulate in the skin of patients while on cysteamine.

There are numerous questions to be answered by this research. For instance, how does cystinosis affect skin pigmentation? What are the connections between the disease and sweating? What are the different ways males and females are impacted?

"We know that cystinosis is associated with accelerated skin aging. Is that something we can measure out of this data?" Dr. Dube asks. "Why do we still see a burden of crystals in the skin of clinical trial participants who have [spent] years on cysteamine and whose leukocyte levels are dose appropriate?"

'WE SO APPRECIATE THE SUPPORT PROVIDED BY THE CRF'

The team is only six months into their project, which has a two-year timeline, so it's too soon to draw conclusions. However, the possibilities drive the researchers to continue exploring with great purpose.

"We are gathering and evaluating data that can affect care now, but perhaps more importantly, it may someday be used to help with diagnosis and cysteamine dosing, to help with thinking about additional treatment options," Dr. Dube says. "That's the hope, that's the aspiration. That's why we so appreciate the support provided by the CRF and the cystinosis community."

There's another hope held by cystinosis patients. Current monitoring of disease progression relies on measuring cystine in white blood cells, necessitating an invasive blood draw. Non-invasive imaging of the skin may give a more accurate estimate of cystine accumulation throughout the body.

As Dr. Dube pursues additional gains from the research project, he says he reaps rewards just by being immersed in the cystinosis community.

"Last year was my first time attending the Day of Hope conference and it was so inspiring not just as a physician and a scientist, but also as a parent of someone who has a rare disease," says Dr. Dube. "It's so energizing to be part of a community with a shared sense of mission."

As his skin imaging project creates exciting new opportunities, Dr. Dube has never felt more invested in advancing the understanding of cystinosis.

"We have made so much research progress," he says, "but we have the hope, desire and energy to do even better."

GETTING THE MOST OUT OF NEW MODELS

by Dennis Arp



AS SHE DEVELOPS NEXT-PHASE MICE TO MEET THIS RESEARCH MOMENT, **DR. VEENITA KHARE** BUILDS ON THE PIONEERING WORK OF HER EXTRAORDINARY MENTOR.

Back in her college days, as she pursued a PhD in cancer biology, Dr. Veenita Khare became fascinated by all the ways that a single-gene mutation could profoundly affect human health. So, she sought out a mentor with a similarly singular focus.

A web search took her to the research page of Dr. Stéphanie Cherqui, whose extraordinary success targeting cystinosis breakthroughs drew Dr. Khare like a tractor beam.

"Stéphanie's translational work provided a prime example to me of bench-to-bedside possibilities, and that was incredibly inspiring," Dr. Khare says.

Now that she is exploring in Dr. Cherqui's internationally prominent research lab at UC San Diego, Dr. Khare cannot imagine another place she would rather be.

"Working with Stéphanie is an incredibly rewarding experience," says Dr. Khare, a post-doctoral fellow in the Department of Pediatrics, UC San Diego Health Sciences. "She's a very supportive mentor who encourages us to ask our own set of questions. She gives us the freedom to explore, but with ultimate goals in mind – to transform understanding and to improve the lives of patients."

MIMICKING THE TRAITS OF CYSTINOSIS

Driven by the lab's spirit of innovation, Dr. Khare has launched a new project full of transformational possibilities. Thanks to grant support from the Cystinosis Research Foundation (CRF), Dr. Khare is developing new mouse models that hold promise as invaluable tools for researchers – colleagues inside the Cherqui lab and others all over the world.

The hope is that these new mouse models will mimic the human phenotype, presenting the characteristics and traits of cystinosis. Such a model would expand exploration of novel treatments based on mutations in the *CTNS* gene that causes cystinosis.

"The more you have a model that reflects what you're seeing in patients, the better it is for research success," Dr. Khare says. "We are fortunate because both of our new models give the same degree of optimism that they will match the phenotype and mechanisms of the disease. If we realize these goals, researchers working on various aspects of the disease can explore different therapy options and perhaps move more quickly toward trials."

In general, animal models play a crucial role in unraveling the pathophysiology of any disease condition, Dr. Khare says.

"Unlike in vitro systems like the cell lines we use for studying a particular disease, animal models provide a more intricate representation of the human aspects of the disease," she explains.

The mouse models for cystinosis developed over the past two decades by the Antignac lab have played an enormous role in the development of global research advancements. For instance, those mouse models are foundational to the groundbreaking stem cell gene therapy currently showing such immense promise in human clinical trials.

A FOCUS ON FANCONI SYNDROME

"We started this study because the initial mouse developed for cystinosis in the Antignac lab has shown genetic drift, with a particular loss of the kidney phenotype," Dr. Khare notes. "This loss has been seen independently across different laboratory settings, and it's especially problematic as we study Fanconi syndrome."

Cystinosis is the leading cause of Fanconi syndrome in children, causing toxic damage to the tubules in kidneys. Despite good treatments like cysteamine, over time, Fanconi can rob the body of nutrients as well as soften bones and cause end-stage renal disease in patients as young as eight.

A key missing element of the current mouse model is that it does not exhibit Fanconi syndrome. With both mouse models being developed in her new project, Dr. Khare has seen Fanconi characterized in mice as young as three months old.

"Our preliminary data show neurobehavioral anomalies along with characteristics of renal Fanconi syndrome," the researcher says. "In addition, cystine storage and cystine crystals, hallmarks of cystinosis, are present in the new models. As we go forward, we plan to explore potential differences between the two models, considering the



Veenita Khare, PhD
Post-Doctoral Fellow, Department of Pediatrics
 UC SAN DIEGO HEALTH SCIENCES

CYSTINOSIS RESEARCH STAR



distinct nature of their mutations and their impact on disease onset or progression."

This project developing new mouse models began with a biotech company and another lab initiating a collaboration. When the host lab closed, the Cherqui lab took over the early research. Dr. Khare notes that developing a new mouse model generally takes one to two years, which is how long she has worked on the project.

CHARACTERIZING THE IMPACT ON ORGANS

Among the important findings of the project so far is that the new mice exhibit behavioral abnormalities, such as impaired spatial perception that mirror those experienced by cystinosis patients.

"I would say that in one more year, we'll be at the point of characterizing the different aspects of the disease and impact on the different organs," she says.

Dr. Khare expects the group will soon be ready to publish results.

"While we are working and compiling data for the manuscript, we have also initiated collaboration with other researchers working in the field," she says. "Obviously, this has already opened a lot of new avenues for us to explore."

The research advances she has enjoyed wouldn't have been possible without the backing of the CRF, Dr. Khare notes. Her journey to success in the Cherqui lab has shown her just how far those avenues can take her when the drivers are collaboration, community, and support.

THE BENEFITS OF A COMMITTED COMMUNITY

The foundation's donors and the grants they make possible are definitely the research engine. But it's the connections the CRF creates that propel the work to breakthrough levels, Dr. Khare enthuses.

"Having a rare life-changing disease like this, it doesn't just affect the person, it affects the whole family," Dr. Khare says.

With cystinosis, an entire supportive community gets involved.

"Because of the foundation, people are able to come from all over the country and the world to meet with each other and with other families who are going through the same thing they are," she says. "You realize you're not alone."

The researchers also benefit.

"I mean, I'm studying about the disease, reading papers and learning so much by meeting other researchers working similarly on the same disease. But maybe even more importantly, I'm meeting patients and their families, seeing how deeply they are involved," Dr. Khare says. "That really motivates you to get everything you can out of your work. Even the smallest possibility might make a big difference."

Elevated by a community that has embraced her, Dr. Khare has become a difference maker in the lab of a world-changer like Dr. Cherqui.

It's everything she dreamed it would be.

"All the projects we're working on interconnect, and we're hoping that these new mouse models will be particularly helpful," she says. "I wouldn't be here if Stéphanie and this community wasn't pushing me. I like working with people who are straightforward and want me to improve. This is all working positively for me." Recently, we also published our work in EMBO Reports, demonstrating how cystinosis regulates the trafficking of a Na⁺/H⁺ exchanger.

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.



Introducing the University of California, Irvine Corneal Cystine Crystal Score: A Novel Tool for Assessing Corneal Crystal Deposition in Cystinosis Patients,
published in ***Scientific Reports***



by Donny W. Suh, MD
Gavin Herbert Eye Institute
University of California, Irvine

scientific reports

This project addresses critical challenges in

cystinosis care, including the difficulty of monitoring disease progression and low patient adherence to treatment. By improving the assessment and treatment of corneal involvement, the project aims to enhance the quality of life for children and young adults with cystinosis. This work will provide clinicians with better tools to monitor the disease and offer more effective, patient-friendly treatment options.

OCTOBER 2025

Co-Induction of Stromal and Epithelial Progenitors for Renal Regeneration, published in
The Innovation: A Cell Press Partner Journal



by Benjamin Freedman, PhD
University of Washington
Seattle, Washington



Dr. Freedman's paper describes how injecting progenitor cells works better than whole organoids. The progenitors can be made from iPS cells in just four days. They contain two different pools of kidney stem cells, which combine to form new filter units. CRF is honored to support Dr. Freedman's groundbreaking research and looks forward to learning more about his progress and the impact it will have as his study advances.

FEBRUARY 2026

A Refined Method for Micro-Scale Blood Cystine Measurement in Preclinical Cystinosis Models, published in the ***International Journal of Molecular Sciences***



by Ester De Leo, PhD

Nephrology Research Unit
Bambino Gesù Children's Hospital
Rome, Italy

Cystinosis and Cellular Energy Failure: Mitochondria at the Crossroads, published in the ***International Journal of Molecular Sciences***



by Francesco Bellomo, PhD

Nephrology Research Unit
Bambino Gesù Children's Hospital
Rome, Italy



International Journal of
Molecular Sciences

Congratulations to Ester De Leo, PhD, and Francesco Bellomo, PhD, of the Nephrology Research Unit at Bambino Gesù Children's Hospital, Rome, Italy, for their recently published papers in the International Journal of Molecular Sciences. These important papers about the discoveries being made in cystine measurement and the role of mitochondria in cystinosis are supporting the development of improved therapies for cystinosis.

FEBRUARY 2026

PFKM Governs Metabolic Shifts Throughout Skeletal Muscle Differentiation, published in ***Nature Metabolism***



by Lauren Albrecht, PhD

Pharmaceutical Sciences
University of California, Irvine

**nature
metabolism**

This study explores how metabolism helps determine cell identity in skeletal muscle. The researchers found that levels of a key enzyme, PFKM, rise as muscle stem cells differentiate, while a signaling pathway (Wnt) can trigger its breakdown through lysosomal degradation. This process shifts how cells use glucose, redirecting it away from energy production toward other pathways that influence cell behavior. In sum, these findings highlight the importance of compartmentalized metabolism in cell fate decisions.

FEBRUARY 2026

Cystinosin Regulates Na⁺/H⁺ Exchanger 3 Trafficking and Function in Kidney Proximal Tubular Cells, published in ***EMBO Reports***



by Stéphanie Cherqui, PhD

Professor, Department of Pediatrics
University of California, San Diego

**EMBO
reports**

Fanconi syndrome is the first clinical manifestation of cystinosis, and it is not prevented by cystine reduction therapy with cysteamine. This study uncovers an evolutionarily conserved novel role of cystinosin in NHE3 trafficking, offering insights into the pathogenesis of Fanconi syndrome and pointing to new treatment possibilities beyond current therapy.

MARCH 2026

Cystinosis Research Foundation is the largest fund provider of grants for cystinosis research in the world.

AUTUMN 2025
\$621,830

2025 TOTAL
\$1,181,024

RESEARCH AND FELLOWSHIP

GRANT AWARDS

CRF has issued 255 grants in 13 countries since 2003.



AUTUMN 2025



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Mentor
Giulia Battaifarano, PhD
Research Fellow
Bambino Gesù Children's Hospital
Rome, Italy

"Exploring the Mechanical Response of Osteogenic Cells in Cystinosis"

\$75,000
ONE-YEAR STUDY



Paul Goodyer, MD
Principal Investigator
Elena Torban, PhD
Co-Principal Investigator
McGill University Health Centre
Quebec, Canada

"mRNA Therapy for Cystinosis"

\$150,000
ONE-YEAR STUDY



Feodor Price, PhD
Principal Investigator
Lee Rubin, PhD
Co-Principal Investigator
Harvard University
Cambridge, Massachusetts

"Characterizing Both the In Vitro and In Vivo Myogenic Potential of Skeletal Muscle Progenitors from Cystinosis Patients"

\$396,830
TWO-YEAR STUDY



Exploring the Mechanical Response of Osteogenic Cells in Cystinosis

Andrea Del Fattore, PhD, *Mentor*

Giulia Battafarano, PhD, *Research Fellow*

BAMBINO GESÙ CHILDREN'S HOSPITAL, ROME, ITALY



OBJECTIVE/RATIONALE:

Patients affected by cystinosis display short stature, osteomalacia, rickets, bone pain and bone deformities. The pathogenesis of bone disorder in cystinosis is multifactorial. Indeed, it is related to renal Fanconi syndrome, and cystinosis deficiency itself contributes to intrinsic bone defect. Additional contributing factors for bone remodeling alteration include malnutrition, copper deficiency, hormonal disturbances and myopathy. In this study, we will explore whether mechanical loading exert beneficial effects on bone cells in cystinosis, and whether it can counteract the adverse effect of cysteamine.

PROJECT DESCRIPTION:

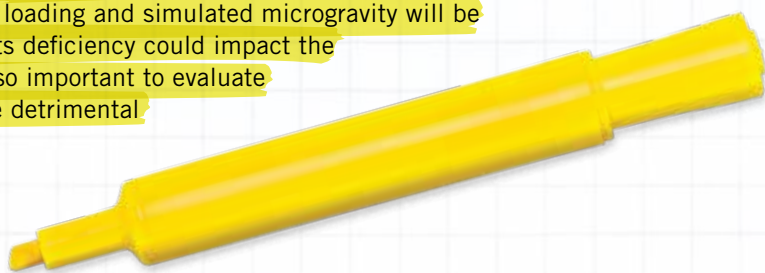
The overall goal of this project is to explore whether mechanical loading may exert beneficial effects on cystinosis lacking (ctns^{-/-}) bone cells and whether it can counteract the adverse effect of cysteamine. To this end, we will perform in vitro experiments aimed at elucidating whether appropriate mechanical loading can improve skeletal function in cystinosis. We will study the direct effect of mechanical unloading and loading on ctns^{-/-} bone cells using NASA-developed Rotary Cell Culture System, a bioreactor to expose cells to altered gravity, and a low-magnitude high-frequency vibration (LMHFV) system, which mimics microgravity and mechanical stimuli, respectively.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

The project has translational impact and could open new insights for the management of bone disease in cystinosis. A better understanding of the effects of mechanical loading on bone cells in cystinosis will be essential to establish whether physical exercise could have a positive or detrimental consequence on bone tissue of patients. It will be important for clinicians to improve patient's quality of life.

ANTICIPATED OUTCOME:

This study will allow us to better understand the role of mechanical loading on bone tissue in cystinosis. In vitro experiments of mechanical loading and simulated microgravity will be essential to discover how physical exercise and its deficiency could impact the skeletal health in patients. The results will be also important to evaluate whether mechanical stimuli could counteract the detrimental effect of cysteamine treatment on bone cells.





mRNA Therapy for Cystinosis

Paul Goodyer, MD, *Principal Investigator*

Elena Torban, PhD, *Co-Principal Investigator*

MCGILL UNIVERSITY HEALTH CENTRE, MONTREAL, QUEBEC

OBJECTIVE/RATIONALE:

When COVID19 arrived, several pharmaceutical companies had already established a technology using small lipid particles (LNPs) to deliver COVID mRNA to our tissues and vaccinate us. We propose to re-purpose this technology as a treatment for cystinosis. Our idea is to inject patients with normal CTNS mRNA in a new Moderna LNP. When taken up by various tissues, the mRNA should generate sufficient normal CTNS protein to accomplish all tasks missing in cystinosis.

PROJECT DESCRIPTION:

Previously, we tested the two forms of CTNS mRNA in cultured human kidney cells. Interestingly, the most abundant form (85%) was especially good at clearing cystine out of lysosomes, whereas the less abundant form (15%) was particularly effective in fixing the system for cleaning up internal cell debris ("autophagy"). We also showed that a novel Moderna LNP could deliver and express mRNA to many tissues of interest in cystinosis (including kidneys, thyroid, and testis).

In this final stage, we will test LNP/mRNA therapy in our new cystinosis mouse model. We will treat mutant mice at an early stage (five months) with six weekly injections of the two CTNS mRNAs combined; this should tell us whether we can prevent the emergence of renal Fanconi. We will then treat mice at a later stage of cystinosis (eight months) to see whether we can reverse established kidney injury.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

Oral cysteamine clear out much of the cystine that accumulates in lysosomes of cystinosis patients. This delays deterioration of kidneys and other organs. However, it is not well-tolerated, and most patients still develop Fanconi Syndrome and need a kidney transplant. By re-purposing Moderna mRNA technology, we should be able to bypass the mutant CTNS gene and deliver normal mRNA that restores all its functions. Although repeated injections would be required, the strategy has the potential to be curative.

ANTICIPATED OUTCOME:

We will ascertain whether early injection of cystinotic mice with LNPs containing normal CTNS mRNA can prevent Fanconi Syndrome and allow repair of established kidney injury. If so, we expect this will prompt major pharmaceutical companies to consider clinical trials of LNP/mRNA in cystinosis.



Characterizing Both the In Vitro and In Vivo Myogenic Potential of Skeletal Muscle Progenitors from Cystinosis Patients

Feodor Price, PhD, *Principal Investigator*

Lee Rubin, PhD, *Co-Principal Investigator*

HARVARD UNIVERSITY, CAMBRIDGE, MASSACHUSETTS

OBJECTIVE/RATIONALE:

Cystinosis patients often suffer from debilitating muscle weakness, but the exact cause remains unclear. This project investigates whether the problem lies within muscle stem cells, which are essential for repairing and regenerating tissue. By using innovative 3D “organoid” models and mouse studies, we aim to pinpoint how these cells fail to function correctly. Ultimately, this work seeks to establish a foundation for new cell-based therapies to restore muscle strength and improve quality of life.

PROJECT DESCRIPTION:

To carry out this project, we will use three main approaches. First, we will grow muscle cells from cystinosis patients in the lab to study how they multiply and form new muscle fibers. Using advanced 3D “organoid” technology, we can create miniature muscle models that better mimic the human body. Second, we will use a cystinosis mouse model to understand how the disease affects natural muscle repair after injury. Finally, we will transplant healthy human muscle stem cells into these mice to see if the cells can successfully restore muscle strength and function. By comparing patient cells with healthy ones, this work aims to prove that cell-based therapies are a viable way to treat the muscle weakness associated with cystinosis.

RELEVANCE TO THE UNDERSTANDING AND/OR TREATMENT OF CYSTINOSIS:

This research will provide a deep understanding of why muscle weakness occurs in cystinosis. By studying muscle stem cells (satellite cells) in 3D “organoids,” we hope to reveal whether the disease stems from an inability of these cells to repair muscle tissue. This project establishes a vital proof-of-concept for using healthy stem cell transplants as a new therapy to restore muscle strength, offering hope for a treatment where none currently exists.

ANTICIPATED OUTCOME:

We expect to uncover the specific cellular malfunctions that cause muscle weakness in cystinosis. By comparing patient-derived muscle cells to healthy ones, the team aims to determine if the disease impairs the ability of stem cells to either multiply or transform into mature muscle fibers. Ultimately, they hope to prove that transplanting healthy, lab-grown muscle stem cells can successfully repair damaged tissue and restore strength, providing a foundation for future human clinical trials.





TO SEE EACH AREA OF RESEARCH FOCUS & GRANTS AWARDED IN DETAIL, VISIT:
WWW.CYSTINOSISRESEARCH.ORG/IMPACT

AREAS OF RESEARCH FOCUS & GRANTS AWARDED SINCE 2003



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—— **10 GRANTS**



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—— **1 GRANT**



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of the Pathogenesis of Cystinosis
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CYSTINOSIS RESEARCH FOUNDATION

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Stéphanie Cherqui, PhD

*Professor, Department of Pediatrics,
Division of Genetics*

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OF DEDICATION TO THE
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The mission of the Cystinosis Research Foundation is to find better treatments and a cure for cystinosis by supporting bench, clinical, and translational research. Since 2003, CRF has raised \$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.

Zoe R. Solsby
Vice President





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