2024 Annual Report

ASSOCIATION FOR CREATINE DEFICIENCIES

Our Vision

Our vision is to have effective treatments and newborn screening for all three cerebral creatine deficiency syndromes (CCDS) while providing community support. In this future, the rare disease diagnostic odyssey changes from seven years to seven days to treatment and all CCDS patients achieve their potential.

Our Mission

To provide patient, family, and public education, to advocate for early intervention through newborn screening, and to promote and fund medical research for treatments and cures for Cerebral Creatine Deficiency Syndromes (CCDS).

Our Story

The Association for Creatine Deficiencies (ACD) was established in 2012, by parents of children diagnosed with Cerebral Creatine Deficiency Syndrome (CCDS) that decided a rare disease needs a unified community to affect change for the future. The ACD Board consists of unpaid CCDS parent professionals committed to this cause.

Executive Director’s 2024 Recap

Dear CCDS Community,

In 2024 we met as a community at our third in-person symposium in Salt Lake City, Utah. The energy was high, the research has been advancing at a quicker pace compared to prior years, and we all left feeling hope. Our first “Champions for a Cure Gala” was held and it featured our CCDS children being introduced as they walked across the stage waving to the audience. What a special night! We launched a second PCORI-funded project, once again centered on THE PATIENT shaping how clinical trials will be designed. Our community grew in numbers - both our patient and research communities!

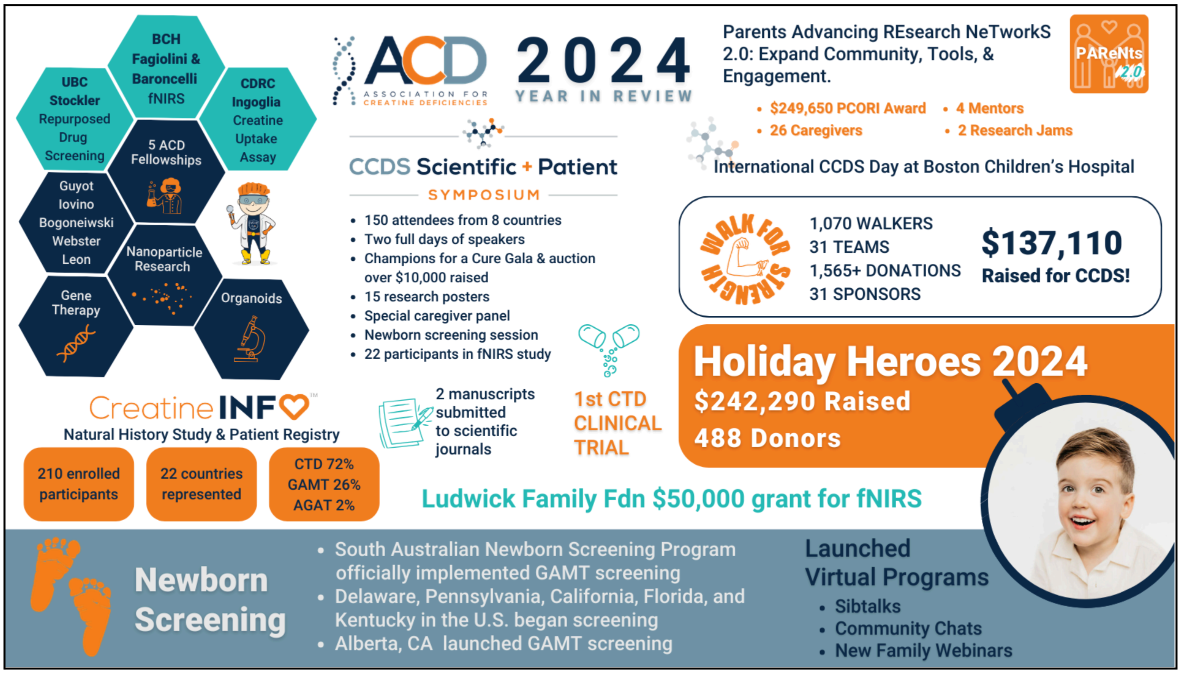
As we conclude this year of major milestones, we look forward with hope and determination. Hope for advancements in therapies under investigation and for quicker diagnoses through newborn screening and growing access to genetic testing. Determination that we will do all we can to push this field forward and find a treatment for CTD, and better treatments for those suffering from late diagnosis of AGAT and GAMT Deficiencies.

Our resolve is strong and we look forward to a year of hard and rewarding work in 2025.

With gratitude and optimism,

Heidi Wallis

Heidi Wallis
ACD Executive Director



ACD 2024 YEAR IN REVIEW

- **UBC Stockler** Repurposed Drug Screening
- **BCH Fagiolini & Baroncelli** fNIRS
- **CDRC Ingoglia** Creatine Uptake Assay
- 5 ACD Fellowships
- Guyot, Iovino, Bogoneiwski, Webster, Leon
- Nanoparticle Research
- Gene Therapy
- Organoids

CreatineINFO™ Natural History Study & Patient Registry

- 210 enrolled participants
- 22 countries represented
- CTD 72%
GAMT 26%
AGAT 2%

CCDS Scientific + Patient SYMPOSIUM

- 150 attendees from 8 countries
- Two full days of speakers
- Champions for a Cure Gala & auction over $10,000 raised
- 15 research posters
- Special caregiver panel
- Newborn screening session
- 22 participants in fNIRS study

2 manuscripts submitted to scientific journals

1st CTD CLINICAL TRIAL

Parents Advancing REsearch NeTworkS 2.0: Expand Community, Tools, & Engagement.

- $249,650 PCORI Award
- 26 Caregivers
- 4 Mentors
- 2 Research Jams

International CCDS Day at Boston Children’s Hospital

1,070 WALKERS
31 TEAMS
1,565+ DONATIONS
31 SPONSORS

$137,110 Raised for CCDS!

Holiday Heroes 2024
$242,290 Raised
488 Donors

Ludwick Family Fdn $50,000 grant for fNIRS

Newborn Screening

- South Australian Newborn Screening Program officially implemented GAMT screening
- Delaware, Pennsylvania, California, Florida, and Kentucky in the U.S. began screening
- Alberta, CA launched GAMT screening

Launched Virtual Programs

- Sibtalks
- Community Chats
- New Family Webinars

Education and Awareness

ACD is the leading international non-profit patient advocacy group raising CCDS education and awareness.

Rare Disease Partners

As part of its commitment to patient and public education, the ACD continues to maintain ongoing relationships with several rare disease and advocacy partners, including:

- The National Organization for Rare Disease (NORD)
- Global Genes
- EveryLife Foundation
- Child Neurology Foundation (CNF)
- ThinkGenetic
- Canadian Organization for Rare Disorders (CORD)
- Southeast Regional Genetics Network (SERN)
- Eurordis
- COMBINEDBrain
- Chan Zuckerberg Initiative (CZI)

Educational Webinars

Sibtalks - Siblings Webinars

Sibtalks is a program designed to help the siblings of children with disabilities have a safe place to share, learn, and process their experiences. Each of ACD's Sibtalks webinars had three separate sessions, divided into age ranges for age-appropriate sibling guidance. Siblings around the world are able to grow friendships through this experience.

Community Chat Series

ACD hosted 3 community chat webinars in 2024. The community chat series is a monthly meeting where the CCDS community hears from experts in various topics in an "ask me anything" style session. View the archive of community chats here.

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Advocacy

ACD is the leading international patient advocacy group that devotes their efforts to changing the lives of people living with all three CCDS. Along with education and awareness, ACD is passionate about advocating for the rights of individuals with CCDS.

Newborn Screening

2024 marked our biggest year yet for newborn screening efforts.

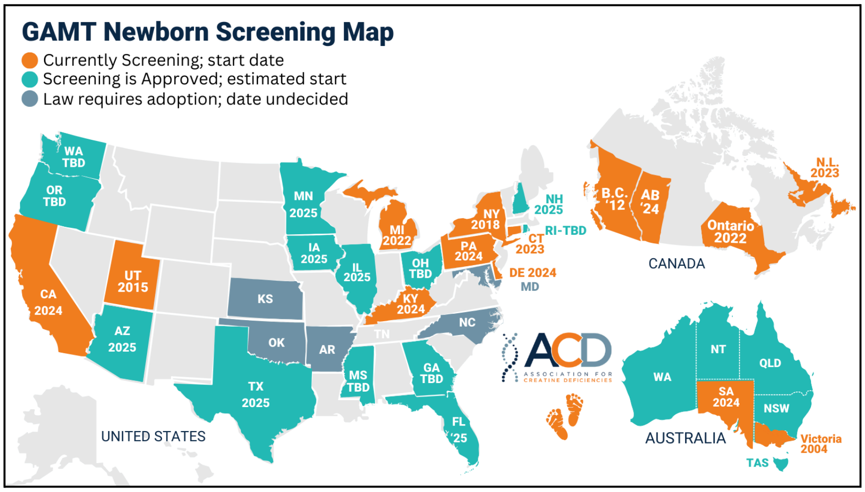
Five U.S. states—California, Kentucky, Pennsylvania, and Delaware—along with Alberta, Canada, and South Australia, Australia, added GAMT to their newborn screening panels.

Additionally, Texas, Minnesota, and Illinois have approved screening and are expected to begin in 2025.

Started screening
California
Kentucky
Pennsylvania
AB - Canada
SA - Australia
Delaware

Screening approved
Texas - est start 2025
Minnesota - est start 2025
Illinois

Law requires adoption
Kansas



Medical and Scientific Research

CreatineINFO Patient Registry

ACD's CreatineINFO Patient Registry & Natural History Study had another successful year in 2024. Some highlights include:

- A 10% growth in new patient enrollment from 2023.
- Partnered with ClinGen's CCDS Variant Curation Expert Panel (VCEP) to support their efforts to reclassify CCDS genetic variants. Using patient-reported data shared in the registry, we successfully reclassified 3 CCDS variants!
- The VCEP published new guidelines for the classification of CCDS genes in the journal "Molecular Genetics and Metabolism". ACD's Executive Director was a contributing author and served as a panel member throughout the year.

- Results from our industry-supported Oral Medication Survey were presented at NORD's annual Rare Diseases & Orphan Products Breakthrough Summit. Check out our poster here!
- Restructured the registry regulatory boards to increase transparency about decision making and to facilitate a more inclusive stakeholder group which better represents our international CCDS community.
- Launched the first CreatineINFO Patient Registry Annual Registry Report. This report communicates key findings from the patient- and caregiver-reported data within the registry. This is a valuable resource for CCDS families to share with their medical team and for researchers to better understand these disorders. We are excited to continue releasing an annual report with more findings each year!

Learn more about the CreatineINFO Patient Registry here.

2024 CCDS Patient + Scientific Symposium, Salt Lake City, Utah

This year, we hosted 150 attendees from 8 countries for a weekend of connection, advocacy, and innovation. This bi-annual, in-person event featured 2 full days of speakers, 15 research posters, a special caregiver panel, a newborn screening session, and 22 participants for an on-site fNIRS study. Our first ever Champions for a Cure Gala & Auction raised over $10,000. Event program and recordings are available here.

Creatine Deficiency Research Center

In the second year of the Creatine Deficiency Research Center (CDRC) at the University of Utah and ARUP Laboratories Dr. Filippo Ingoglia began work on a creatine uptake assay for CTD. The goal of this project is to study variants of *slc6a8,* provide diagnostic and reclassification support, and increase variant understanding to support caregivers and clinicians in identifying variants with (or without) residual transport activity - informing decisions around supplementation potential.

2024 ACD Fellowship Grants

- Sebastian Leon received the award to pursue research to establish that creatine-loaded nanoparticles can pass the blood-brain barrier. Leon is working under the mentorship of Dr. Swadesh Santra at the University of Central Florida.
- Troy Webster will conduct preclinical dose-finding studies on a gene therapy for CTD. Webster is supported by mentor Dr. Jagdeep Walia at Queen's University in Kingston, Ontario, Canada, who is overseeing an ongoing CTD gene therapy project which Webster's fellowship project will support.
- Anne-Cecil Guyot is pursuing an organoid project under mentor Dr. Aloise Mabondzo at CEA/Paris Saclay University in Paris. This project will focus on using human brain organoids from CTD patients to see if a correction can be triggered in target proteins in response to pharmacological interventions.

- Ludovica Lovino, Ph.D., will further efforts on gene therapy investigation for CTD in the lab of Dr. Laura Baroncelli at the Institute of Neuroscience in collaboration with Fondazione IRCCS Stella Maris, both in Pisa, Italy.
- Aleksander Bogoneiwski will characterize CTD and Guanidinoacetate Methyltransferase (GAMT) Deficiency patient cell-derived organoids to find effective gene therapies under the guidance of Dr. Gerry Lipshutz at UCLA.

PAReNts 2.0: ExCiTE Project

After the successful development of a Core Outcome Set (COS) for CTD and GAMT Deficiency at the beginning of 2024 (you can view the preprint of the paper describing this work here), ACD was awarded funding for a second project - Parents Advancing REsearch NeTworkS 2.0: Expand Community, Tools, & Engagement (PAReNts 2.0: ExCiTE). The project kicked off in September and a new group of 24 caregivers joined the effort to study research engagement in monthly workshops. Our co-PI on this project is Dr. Audrey Thurm and the focus of the project is establishing the smallest meaningful differences parents hope for in a successful clinical trial, specifically in the areas of the Core Outcome Set. Additionally, considerations for the appropriate tools to measure these small changes are to be created as a companion to properly using the COS in a clinical trial.

CCDS Caregiver & Patient Community

ACD places a strong emphasis on community. Through our community engagement programs, we believe in fostering strong personal ties to those patients and families living with CCDS so that collectively we can become better caregivers, advocates and champions for our children, and each other.

2024 Walk for Strength

The 2024 Walk for Strength had 1,070 participants on 31 teams. This walk serves as ACD's signature awareness and fundraising event and is a great way to get involved, no matter the size of your contribution. There were more than 1,565 donors and 31 sponsors, and together, we raised $137,110!

Thank you to our Supporter and Booster level sponsors:

Victory Taekwondo
Big Sky Oral & Facial Surgery
Rusty Taco
Vulcan Materials Company
Brighton Park Capital

CCDS Day - February 1, 2024

Our goal this year was to spread awareness about CCDS and provide meaningful connections for newly diagnosed CCDS families through our campaign - "This is CCDS - Raising Awareness and Supporting Families".

We invited CCDS caregivers to submit short videos offering advice and encouragement to families facing a new diagnosis. These heartfelt messages were compiled into a special video for CCDS Day. Watch the video here.

Additionally, we celebrated CCDS Day with a hybrid event in collaboration with Boston Children's Hospital. Eleven speakers presented on the latest CCDS research. Watch all 11 presentations here.

CCDS Patient Growth

In 2024, the ACD Family Network grew 12% with families from 17 countries finding us. It is wonderful to see so many new families connecting with ACD to receive important information, resources, and support. We continue to welcome new families, connecting them with other CCDS families in their region, and encouraging them to engage with the established CCDS and GAMT support groups online. We also invite new families to join the CreatineINFO Patient Registry.

Fundraising

The ACD is a 501(c)(3) charitable non-profit organization which can provide valuable services to the CCDS community only because of the generosity of its contributors. Special Events and Corporate Giving are essential to raising CCDS awareness and funds needed to continue the ACD's mission.

Holiday Heroes 2024

Holiday Heroes is ACD's longest running program. Started in our first year, we reach out to our community, friends, families, and other supporters to raise funds that support the following year's research efforts. The focus of our 2024 Holiday Heroes campaign was "Cure Creatine Deficiencies/Give for Research".

2024 was a very successful Holiday Heroes campaign. Thirty-four teams worked together, and 488 generous donors—including one anonymous $50,000 contribution—helped us raise a total of $242,290 for research.

These funds will support an active research program in 2025, including five research fellowships.

ACD Board

ACD was founded in 2012 by parents with children diagnosed with a CCDS. ACD was established to raise awareness and education of CCDS among the medical community and the general public, and to advocate on behalf of the families and patients living with CCDS.

ACD aims to bring the CCDS community together as one strong voice to promote newborn screening and medical treatments and, one day, cures for CCDS.

Together, we are "Building Strength and Changing Lives."

2024 Board of Trustees:

Dan Coller
Board Chair
Kim Tuminello
Director of Advocacy
Erin Coller
Director of Communications

Jeffrey Randall Allen
Vice Chair, Director of Impact & Donor Relations
Celeste Graham
Director of Education
Mikelle Law
Director of Finance

2024 ACD Scientific Medical Advisory Board:

Dr. Nicola Longo, M.D., Ph.D.
Dr. Saadet Andrews, M.D., Ph.D., FCCMG, FRCPC
Dr. Andreas Schulze, M.D., Ph.D., FRCPC
Dr. Sylvia Stockler, M.D.

Dr. Marzia Pasquali, Ph.D., FACMG
Dr. Sarah Young, Ph.D.
Dr. Ton DeGrauw, M.D., Ph.D.
Dr. Gajja Salomons, Ph.D.
Dr. Matthew Skelton, Ph.D.

ACD 2024 Financial Accounting

Statement of Financial Position
As of December 31st, 2024

Liabilities & Net Assets

Net Assets, Beginning of Period	$ 647,957
Change in Net Assets	- 239,712
Net Assets End of Period*	**408,245**

***Net Assets Include**

Assets With Donor Restrictions	12,825
Assets Without Donor Restrictions	$395,420

Statement of Activities
As of December 31st, 2024

Ordinary Income/Expense

Income	
Grants and Contributions	$ 235,105
Contributed Services	24,593
Special Event Revenue	118,766
Investment Income	7,552
Conference Revenue	47,133
Other	5,576
Total Income	**438,725**
Expense	
Program Services	536,689
General and Administrative	94,400
Fundraising	47,348
Total Expense	**678,437**
Change in Net Assets	**-$239,712**

2024 Audit and Form 990 prepared by: Mullins P.C., Certified Public Accountants