2023 Annual Report

ACD

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 2023 Recap

Dear CCDS Community,

We are grateful for the amazing progress which was made in 2023 thanks to the support of our patient and scientific communities. This year was a historic one for our rare disease community. The once-in-a-lifetime milestones met in 2023 include:

1. GAMT Deficiency was officially added to the U.S. Recommended Uniform Screening Panel. This milestone was reached nearly seven years after the first nomination of GAMT for RUSP inclusion and after MANY presentations by ACD advocates and scientists committed to seeing this disorder diagnosed at birth and changing the lives of future generations.

2. ACD hosted the CCDS Externally-Led Patient-Focused Drug Development (EL-PFDD) Meeting. This meeting is a crucial milestone for creating an accessible account of the struggles and hopes of the CCDS patient community for the awareness of policymakers, scientists, and industry leaders involved in care and treatment development for AGAT, GAMT, and CTD.

3. The Voice of the Patient (VOP) report is the official documentation of the EL-PFDD Meeting. This is yet another one-time-only milestone met in 2023. The VOP is distributed far and wide and is posted on the FDA website in addition to ACD's as a perpetual record of the experience of living with a creatine deficiency.

4. ACD's first PCORI grant concluded at the end of 2023 with a Consensus Meeting that assembled the data from the two-year study along with a consensus group which easily agreed upon the eight core outcomes that should be measured in all future clinical trials for GAMT and CTD.

5. The first-ever Creatine Deficiency Research Center launched in March of 2023 signalling the ACD's commitment to long-term support of research efforts and partnership as well as the patient community's willingness to donate for key causes.

We could not have completed these important initiatives without the support of our families, scientists, donors, and others that are committed to this cause.

With gratitude and hope,

Heidi Wallis

Heidi Wallis
ACD Executive Director

Education and Awareness

ACD is the only 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

Sibshops - Siblings Webinars

Sibshops 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 Sibshops 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.

CCDS Expert Panel Community Webinar

ACD hosted 3 Expert Panel webinars in 2023. These virtual panel events include the world's leading CCDS experts to discuss new approaches toward common struggles faced by parents with AGAT, GAMT, and CTD. These regular events keep researchers acutely aware of the patient perspective and allow families to ask questions. Topics of discussion in 2023 included seizures, behavioral issues, speech and communication, gastrointestinal issues, cardiovascular concerns, sleep disturbances, and supplements.

Externally-Led Patient Focused Drug Development (EL-PFDD)

ACD hosted a virtual Externally-Led Patient Focused Drug Development (EL-PFDD) meeting on CCCS on January 24, 2023. This meeting provided an important opportunity for patients and caregivers to inform FDA representatives, academic and scientific researchers, medical professionals, and pharmaceutical companies about personal experiences regarding the symptoms and daily impact of CCDS, as well as thoughts on current and future approaches to therapies. The meeting was attended by 248 individuals, including CCDS patients, caregivers, family, friends, healthcare providers, government and industry representatives, and research scientists. More information about the EL-PFDD meeting can be found here.

Voice of the Patient Report

The CCDS Voice of the Patient Report is an impactful summary of the EL-PFDD meeting with many quotes from caregivers and live polling results presented in an insightful manner. ACD has provided this report to the U.S. Food and Drug Administration, government agencies, regulatory authorities, medical product developers, academics, and clinicians, and it is publicly available for the many stakeholders in the CCDS community.

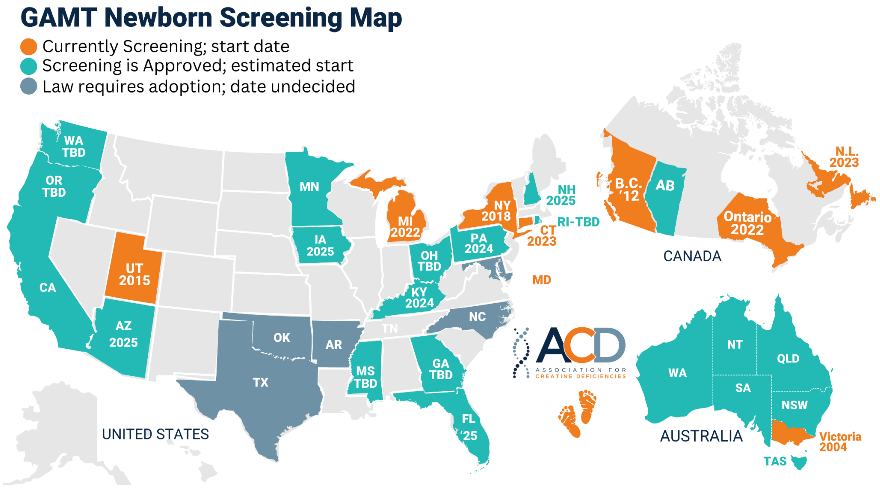
We invite you to read the Voice of the Patient Report and learn more about the CCDS community.

Advocacy

ACD is the only 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

The U.S. Secretary of Health officially approved GAMT for addition to the Recommended Uniform Screening Panel on January 23, 2023. Connecticut in the US and the Newfoundland and Labrador province of Canada added GAMT to their newborn screening panels in 2023.



Medical and Scientific Research

CreatineINFO Patient Registry

ACD's CreatineINFO Patient Registry & Natural History Study had a successful 2023. It reached an enrollment of over 200 CCDS participants from around the world. It launched a Cardiovascular Health survey in partnership with NIH pediatric cardiologist, Dr. Mark Levin. It also released the Oral Medication Preferences survey, which was developed in partnership with Ultragenyx. This marks the first custom registry survey developed in collaboration with an industry partner.
Learn more about the CreatineINFO Patient Registry here.

2023 CCDS Virtual Conference

The focus of this conference was to share the latest CCDS research and expand our research network. We heard from researchers on a variety of topics and were joined by industry partners who provided insights into therapeutic development for rare diseases.
ACD hosted 34 speakers across 9 sessions. Caregivers, researchers, scientists, medical professionals, and industry stakeholders attended and participated in Q&A discussions following each session. View talk recordings and find speaker abstracts and bios here.

Creatine Deficiency Research Center

In 2023, ACD launched the first Creatine Deficiency Research Center (CDRC) at the University of Utah and ARUP Laboratories. The first project funded through this collaboration was led by Dr. Steven Baker and focused on exploring a gene therapy treatment for CTD. Additional focus at the CDRC is on exploring potential for a newborn screening assay for CTD and a creatine uptake assay for CTD diagnostic support and understanding, and reclassification support, for various slc6a8 genetic mutations.

2023 ACD Fellowship Grants

Three young researchers were the recipients of an ACD Fellowship grant in 2023:

- **Ludovica Iovino**, in the lab of Dr. Laura Baroncelli focused her research on the development of gene therapy as a possible treatment for Creatine Transporter Deficiency.
- **Crystal Mulik**, in the lab of Dr. Saadet Andrews was selected for her proposal to advance biomarker discovery and development of a zebrafish model of CTD for drug screening.
- **Jacklyn Gallagher** received a 2023 ACD fellowship to pursue research activities in the lab of Dr. Jonathan Schlebach. Her work focused on pharmacological chaperones for Creatine Transporter Deficiency.

PAReNts: Core Outcome Set Development

We successfully completed the PCORI-funded Parents Advancing REsearch NeTworkS (PAReNts) Project this year. The goal of this project was to engage, train, and empower a group of CCDS caregivers to collaborate with clinicians and researchers to advance CCDS research. A special focus of this project was the development of a Core Outcome Set (COS) for CTD and GAMT deficiency that accurately represents the most important outcomes that should be measured in every clinical trial. In November, 25 CTD and GAMT caregivers, clinicians, and researchers gathered in Salt Lake City, Utah, USA at a consensus workshop to finalize the COS for CTD and GAMT deficiency.

Read our report on this final list of 8 outcomes in the COS for CTD and GAMT deficiency here.

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.

2023 Walk for Strength

The 2023 Walk for Strength had 1,115 participants on 34 teams across 8 countries. 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 321 donors and 30 sponsors, and together, we raised $92,264!

CCDS Day- February 1, 2023

Families participated in ACD's CCDS Day Campaign, "Learn | See | Screen," with the goal of spreading the word that physicians need to screen for creatine deficiencies and that concerned parents should request screening. A companion educational video was created in 8 different languages for CCDS 2023. View the playlist here. ACD raised awareness about the importance of screening through social media.

CCDS Patient Growth

In 2023, the ACD Family Network experienced a 19% growth in families from more than 20 countries. 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 2023

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 2023 Holiday Heroes campaign was "Give Hope". 2023 was a very successful Holiday Heroes campaign, with over $201,067 raised by nearly 491 donors. These funds will support an active research program in 2024 including continuation of research projects at the CDRC at the University of Utah.

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"

2023 Board of Trustees:

2023 ACD Scientific Medical Advisory Board:

ACD 2023 Financial Accounting

Statement of Financials
January to December 2023
Balance

Liabilities & Net Assets

Net Assets, Beginning of Period	$ 659,951
Net Income	- $ 11,994
Net Assets*	**$647,957**

*Net Assets Include

Total Assets, End of Period	$ 674,679
Assets With Donor Restrictions	$ 167,357
Assets Without Donor Restrictions	$ 480,600
Total Liabilities	- $ 26,722

Statement of Financials
January 1 through December 31, 2023
Balance

Ordinary Income/Expense

Income

Grants and Contributions	$ 534,004
Contributed Services	$ 19,616
Special Event Revenue	$ 45,130
Investment Income	$ 2,487
Other	$ 6,571
Total Income:	**$607,808**

Expense

Program Services	$ 493,631
General and Administrative	$ 69,740
Fundraising	$ 39,242
Total Expense	**$ 622,229**
Net Income	**-$14,421**

2023 Audit and Form 990 prepared by: Mullins P.C., Certified Public Accountants