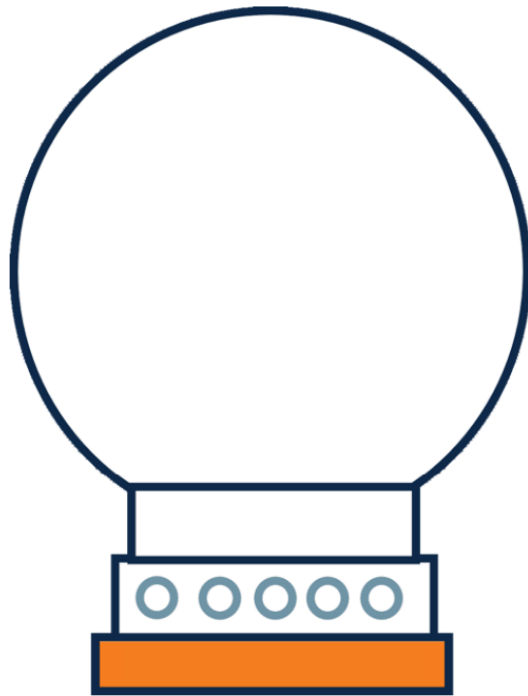


2021 Annual Report





Our Vision

Our vision is to have effective treatments and newborn screening for all three 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.

President's Letter

Dear CCDS Stakeholders,

2021 was a record-breaking year. We awarded more than \$200,000 toward research fellows, drug screening projects, gene therapy grants, and similar projects. GAMT Deficiency was approved by the US Advisory Committee on Heritable Disorders in Newborns and Children for a full evidence review. We launched the CreatinineInfo Patient Registry and nearly half of our community has joined the registry so far. We were awarded, and began work on, a PCORI grant that will help us define core outcomes, shaping future clinical drug trials. These are only a few of the "big wins" for CCDS this past year.

All of this is thanks to an active community of supporters giving their time and money to make these advancements possible. We are beyond grateful for all who have given their support and look forward to an even more exciting 2022.

A handwritten signature in black ink, reading "Heidi Wallis". The signature is fluid and cursive, with the first name "Heidi" and last name "Wallis" clearly distinguishable.

Heidi Wallis

ACD President

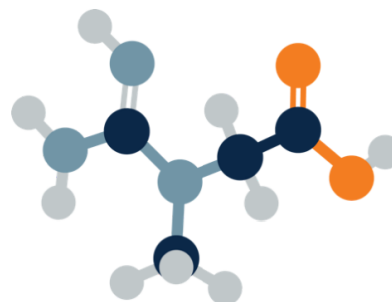
Education and Awareness

The ACD is the only 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
- Baebies
- Child Neurology Foundation (CNF)
- ThinkGenetic
- Canadian Organization for Rare Disorders (CORD)



Website and Social Media

ACD continues to utilize the website creatineinfo.org and our social media channels to increase educational and key messaging.

12 blogs were shared on the ACD [blog](#) with research information and CCDS family stories, reaching the newly diagnosed and extending the ACD community internationally.

CCDS Day and Rare Disease Day

CCDS Day (Feb. 1) and Rare Disease Day (Feb. 29) continue to be important days for the CCDS community to unite under our shared cause. These awareness days bring critical attention to GAMT, AGAT and CTD. It is important to continue participating in awareness days to give these rare disorders a louder voice. We've seen a huge increase in participation and are excited that these days are receiving global recognition.

Advocacy

ACD is the only 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 United States Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) voted unanimously to advance Guanidinoacetate Methyltransferase Deficiency (GAMT) forward for review by the Evidence Review Committee to add GAMT to the Recommended Uniform Screening Panel for all newborn babies.

Advocacy Efforts

Rare Across America Virtual Event: Advocates met virtually with senators and representatives to participate in advocacy events.

Kim Tuminello, ACD's Director of Advocacy, attended a virtual meeting with CA Congressman Mike Levin to gain support on the following bills: The Reauthorization of the Newborn Screening Saves Lives Act, The STAT Act and The Access to Genetic Counselor Services Act.

The ACD community was called upon to advocate for protection of the Orphan Drug Tax Credit (ODTC), which allows rare disease patients to have access to an FDA approved drug for their condition.

Medical and Scientific Research

2021 was a busy year in promoting research efforts and developing the CCDS Patient Registry for the Association for Creatine Deficiencies.

ACD awarded three fellowships, funded gene therapy projects, and produced four educational webinars. ACD launched the Registry for Creatine Deficiencies on March 15, 2021.

Research Meetings

2021 CCDS Virtual Conference: ACD organized a virtual conference that brought together researchers, scientists, medical professionals, industry stakeholders, and CCDS families with the goal of sharing the latest CCDS research and to broaden ACD's research network. Invited speakers covered a wide range of topics.

Research Grants

The ACD awarded 3 research fellowship grants, funded gene therapy research and funded additional screening for inhibitors in guanidinoacetate synthesis.

\$25,000 Award - Dr. Nicola Longo

This award will go towards the discovery of an inhibitor of the synthesis of guanidinoacetate. The goal is to find a potential drug which, given with creatine supplements, can normalize the brain chemistry of GAMT patients, thus reducing the frequency of seizures and facilitating intellectual development. The current grant will be used for screening potential inhibitors in cells.

Gene Therapy Advancement Award - \$10,000

Awarded to Dr. Olivier Braissant, Ph.D.

ACD awarded the GTA grant to Braissant to enable testing of viral vector mediated gene therapy in a novel rat model of Creatine Transporter Deficiency (CTD). With this award, Braissant's lab will be able to test a viral vector to treat CTD in mutant rats, measure correction of creatine levels in the brain and determine if behavioral issues can also be improved by this method of gene therapy. The study also aims to provide insights to help with choosing the best applicable protocol to be used in humans. For instance, Braissant will look at identifying the best delivery route, best dose of AAV vectors, and best time of delivery.

\$30,000 Fellowship Award - Dr. Charles Kuntz in the lab of Dr. Jonathan Schlebach

Dr. Kuntz and Dr. Schlebach are conducting virtual screening of different patient gene mutations. This work is expected to shed light on the functional impact of patient mutations and explores potential avenues for therapeutic rescue.

\$30,000 Fellowship Award - Dr. Peter Axerio-Cilies in the lab of Dr. Sylvia Stockler

Dr. Stockler and Dr. Axerio-Cilies will work on screening compound libraries on cells with various patient mutations, in keeping with ACD's mission to advance translational therapies.

\$30,000 Fellowship Award - Alex Lee in the lab of Dr. Andreas Schulze

Alex Lee intends to work on understanding regulatory mechanisms for genes critical to the creatine synthesis and transport pathway.

Educational Webinars

The Rare Sibling Experience

A conversation on commonly observed barriers and best practices for fostering healthy relationships between rare children and their siblings. This webinar was hosted by Kate McGowen, Child Life Specialist and Dr. David Rintell, Psychologist.

Rare on the Road

A tutorial on how to share their rare disease story and get involved in advocacy. This webinar was hosted by Global Genes and EveryLife Foundation.

How to Read Your Genetic Test Report

An educational webinar designed to make families more active participants in finding treatments for their loved ones, by promoting a full understanding of the diagnosis. This webinar was hosted in partnership with ClinGen.

The Clinical Trials Process

A deeper dive into understanding the clinical trials process, hosted by Dr. Sheila Farrell, MD, MPH from the FDA's Center for Drug Evaluation and Research.

Patient and Family Supportive Services

ACD places a strong emphasis on patient and family support. The need for resources and community are essential in both navigating and living with a rare disease. 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.

2021 Walk for Strength

The 2021 Walk for Strength had 636 walkers in 7 countries. This walk serves as a signature awareness and fundraising event for ACD and is a great way to get involved, no matter how big or small. There were 260 donations and 22 sponsors. Together we raised \$78,522.

CCDS Day Campaign

Families participated in ACD's CCDS Day Campaign, "Share your CCDS Story" as each family shared their challenges, hopes and the impact of CCDS on their family. Families used the hashtag #MyCreatineStory on their social media accounts and family stories were highlighted on ACD's social media accounts. CCDS Day continues to raise awareness and serves as an opportunity to unite the three disorders together for a shared cause.

CCDS Patient Growth

In 2021, the ACD Family Network grew from 232 to 272 families from more than 30 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 with welcome packets containing CCDS Educational materials and we encourage them to find others online in the established CCDS and GAMT support groups.

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 our contributors.

Special Events and Corporate giving are essential to raising CCDS awareness and funds needed to continue the ACD's mission.

The following is a list of areas of sources, which provided funding for 2021:

Contributors

Sponsors:

Ultragenyx
Advanced Corrosion Technologies & Training (ACTT)
Big Sky Oral & Facial Surgery
Marten Transport
CZI
Fitz Roofing
Sullivan Ranch
Meredith Contracting
International Brotherhood of Electrical Workers (IBEW)
2 P's Ranch
King & Co Real Estate

Trumpet Behavioral Health
Triwest
Buggy Bath Car Wash
Ceres Brain Therapeutics
Alzchem
Edelife
Therapy Talk

Special Events:

AmazonSmile
Prime Day
2021 Walk for Strength
Holiday Heroes

Grants Received

Eugene Washington Engagement Award by the Patient-Centered Outcomes Research Institute (PCORI), supporting Year 1 of the PaReNts Project.

Chan Zuckerberg Initiative, Rare as One Capacity Building Award, Year 2.

We gratefully acknowledge these generous donors, each playing a vital role, in carrying out our mission to serve CCDS patients and families.

And to all our donors, sponsors and volunteers across the country who helped our various events, it is with sincere gratitude we thank you. Thank you for your continued support and generosity.

Our 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.”

2021 Board of Trustees:

Heidi Wallis

Board President, Trustee

Laura Trutoiu

Vice President, Trustee

Kim Tuminello

Director of Advocacy, Trustee

Erin Collier

Director of Communications, Trustee

Dave Collier

Director of Finance, Trustee

Mikelle Law

Director of Compliance, Trustee

Randy Allen

Director of Impact and Donor Relations,
Trustee

2021 ACD Scientific Medical Advisory Board:

Dr. Nicola Longo, M.D., Ph.D.

Dr. Bruce Barshop, 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. Sarah Young, Ph.D.

Dr. Denise Morita, M.D.

Dr. Ton DeGrauw, M.D., Ph.D.

Dr. Yiumo Chan, Ph. D

Dr. Gajja Salomons, Ph. D.

Dr. Matthew Skelton, Ph.D.

ACD 2021 Financial Accounting

Statement of Financials January to December 2021 Balance

Liabilities & Net Assets

Net Assets, Beginning of Period	\$706,433
Net Income	- \$141, 535
Net Assets*	\$564,897

*Net Assets Include

Total Assets, End of Period	\$574,512
Assets With Donor Restrictions	\$ 249,450
Assets Without Donor Restrictions	\$ 315,447
Total Liabilities	- \$ 9,615

Statement of Financials
January 1 through December 31, 2021
Balance

Ordinary Income/Expense

Income

Grants and Contributions	\$ 285,982
Contributed Services	\$ 18,000
Special Event Revenue	\$ 34,760
Investment Income	\$ 10,246
Other	\$ 1,531

Total Income: **\$ 350,519**

Expense

Program Services	\$ 414,560
General and Administrative	\$ 56,602
Fundraising	\$ 20,892

Total Expense **\$492,054**

Net Income **- \$141, 035**