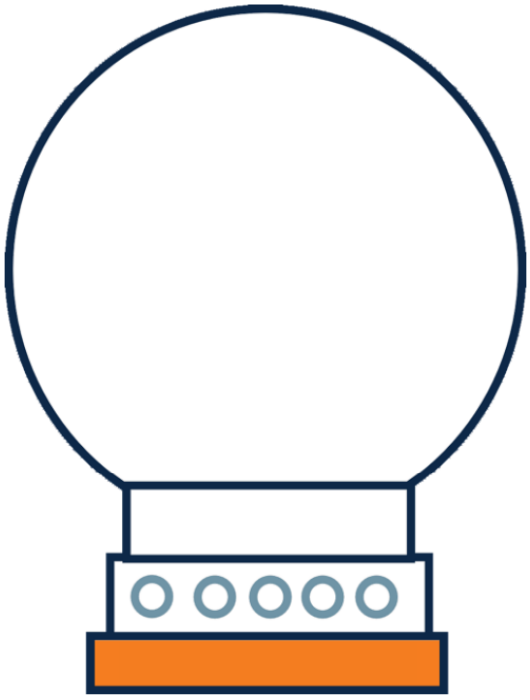
2022 Annual Report

ACD
ASSOCIATION FOR
CREATINE DEFICIENCIES



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.

Executive Director's 2022 Recap

Dear CCDS Community,
It is with great excitement that we conclude 2022 and look towards 2023. Over the past two months, our community rallied and raised over $255,000 as part of our Holiday Heroes 2022 campaign. Thanks to your support we are starting 2023 strong with ambitious goals including the launch of a Creatine Deficiency Research Center with several research projects set to roll out in 2022 and beyond. Here are a few additional highlights from 2022:

- After 7+ years of advocacy, GAMT Deficiency was recommended for inclusion in the RUSP in 2022 and awaits only the U.S. Secretary of Health and Human Services' signature to make it official.
- We completed our 3rd and final year as Rare As One recipients of Chan Zuckerberg Initiative funding to build capacity and as part of that capacity, we hired our first Executive Director, giving our organization full-time leadership to better support our board's success and our mission-driven initiatives.
- ACD obtained funding and made preparations in 2022 to host the CCDS Externally Led Patient-Focused Drug Development (ELPFDD) meeting in January 2023. We look forward to engaging with our community, clinicians, and the FDA to raise awareness and understanding of CCDS and to produce the Voice of the Patient Report in 2023.
- The second CCDS Scientific + Patient Symposium took place in June of 2022 in Park City, Utah.
- Our PCORI funded project "Parents Advancing Research Networks" (PaReNts) kicked off in 2022 with the recruitment of 24 parent participants in the spring, a focus group held at the symposium, and monthly training sessions with our research team in partnership with Dr. Sylvia Stockler and Dr. Audrey Thurm.
- We hosted our first ever CCDS Expert Panel meeting s part of the PaReNts project, bringing together clinical experts to discuss patient cases in the context of approaches to improving care.
- 4 researchers completed work as 2022 ACD Fellowship awardees.
- ACD received a grant to support development of ICD10 codes for CCDS in 2023.
- 2022 WFS was our biggest walk yet, raising $88,489!

Thank you for coming alongside us. Here's to a successful 2023 and major advances in CCDS research!

Heidi Wallis
ACD Executive Director

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
- Child Neurology Foundation (CNF)
- ThinkGenetic
- Canadian Organization for Rare Disorders (CORD)
- Southeast Regional Genetics Network (SERN)

Website and Social Media

ACD continues to utilize the website creatineinfo.org and our social media channels to increase educational and key messaging. Over 42,000 people were reached through Facebook and Instagram, and there were 19.300 visitors to the ACD website.

4 blogs were shared on the ACD blog with research information and CCDS family stories, reaching the newly diagnosed and extending the ACD community internationally. CTD dads Randy Allen & Nate Vandenberg launched a new podcast “Ramblin’ Rare” on all major podcast platforms.

CCDS Day and Rare Disease Day

CCDS Day (Feb. 1) and Rare Disease Day (Feb. 28) 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

Ontario, Canada and Michigan, USA added GAMT to their newborn screening panel.
Universal GAMT newborn screening was advanced for approval by the U.S. Secretary of Health.
The first baby in Australia with GAMT found through newborn screening was diagnosed.

Advocacy Efforts

Virtual Rare Disease Week on Capitol Hill brought together rare disease advocates to make their voices heard by Members of Congress. Participants learned about policies impacting rare disease advocacy and directly advocated for policy change in meetings with Members of Congress.

ACD Challenge: Families shared their CCDS stories across many platforms, but most widely shared in an article in USA Today.

Helping Experts Accelerate Rare Treatments (HEART) Act was supported by Senator Casey, PA, and Senator Scott, SC. ACD families were asked to ask their senators to co=sponsor the HEART Act.

Patient Registry Advocacy: ACD joined 41 rare disease organizations to advocate for patient registries and suggested ways regulators, patient organizations and industry can work together to leverage the potential of real-world data (RWD) and translate it into real-world evidence (RWE).

Coriell Institute: CCDS families are encouraged to submit biosamples from their affected family members to ensure their genetic variant is being represented in the current Coriell Institute research and any research projects that utilize Coriell specimens.

Creatine Info Patient Registry

Following a successful launch in March 2021, the CreatineInfo patient registry and natural history study continues to grow with 156 participants currently enrolled, representing 20 countries.

Medical and Scientific Research

2022 was a busy year in promoting research efforts and developing the CCDS Patient Registry for the Association for Creatine Deficiencies.

ACD awarded four fellowship awards, granted one gene therapy award, executed one contract research project, and produced five educational webinars or webinar series.

Research Meetings

CCDS Scientific + Patient Symposium: Connecting for a Cure was a huge success. There were 7 sessions and 27 speakers over two days in Park City, UT. You can view talk recordings and find speaker abstracts and bios here.

Research Grants

ACD Gene Therapy Advancement Award - $10,000

The GAMT Gene Therapy Project of Dr. Gerald Lipshutz of UCLA was funded by ACD for the second time, after promising initial results. In a mouse model of GAMT deficiency, they found that creatine levels normalized, as did brain activity and learning in the mice. The second round of funding will support a project to determine the strength of dose needed to achieve these results.

$30,000 Fellowship Award - Dr. Charles Kuntz in the lab of Dr. Jonathan Schlebach
$30,000 Fellowship Award - Dr. Peter Axerio-Cilies in the lab of Dr. Sylvia Stockler

Dr. Kuntz and Dr. Schlebach will work with Dr. Axerio-Cilies and Dr. Stockler to identify drugs that may rescue creatine uptake across different types of SLC658 mutations. These fellowship projects compliment each other in characterizing specific mutations and how they respond both in computer simulations and petri dish experiments to existing compounds.

Through these studies, research is identifying potential treatments targeting specific genetic variants by identifying repurposable drugs that can restore functional activity (creatine uptake) of the dysfunctional SLC6A8 transporters. Results from the first year of these studies showed that there are existing compounds that may increase creatine transport in non-functional mutations.

$30,000 Fellowship Award - Léa Broca-Brisson in collaboration with Dr. Aloise Mabondzo

Léa Broca-Brisson is focused on development of human brain organoids from CTD patients as a tool for evaluating the efficacy of potential treatments. She plans to identify biomarkers as a standard for measuring treatment success, particularly in relation to cognitive improvements.

$15,000 Fellowship Award - Alex Lee in the lab of Dr. Andreas Schulze

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

Educational Webinars

Coriell Biobanking Webinar

Families were encouraged to participate in Coriell research and learn about the importance of biobanking in advancing research.

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 the regularly scheduled webinars had three separate sessions, divided into age ranges for age-appropriate sibling guidance.

Virtual Town Hall with Ultragenyx: Updates on the Vigilan Study

The principal investigators of the Vigilan Natural History study met with parents and families of individuals with CTD who contributed to the study. They answered questions and announced that they closed the study ahead of schedule, which meant the study's manuscript and analysis was able to be published early.

CCDS Expert Panel Community Webinar

ACD launched the Expert Panel, a virtual panel event composed of 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. Patients were advised to invite their doctor.

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

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 CCDS Voice of the Patient Report is an impactful summary of this meeting with many quotes from caregivers and live polling results presented in an insightful manner. We invite you to read this report and learn more about our community.

Drug Development and Clinical Trials Process Webinar

Dr. Shiela Farrell of the FDA's Center for Drug Evaluation and Research (CDER) taught families about the clinical trials process, an important process for CCDS families to understand as trial opportunities arise.

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.

2022 Walk for Strength

The 2022 Walk for Strength had 1,189 walkers on 50 teams in 12 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 347 donations and 38 sponsors. Together we raised $88,489!

CCDS Day Campaign

Families participated in ACD's CCDS Day Campaign, “CCDS Research Success Means…” as each family shared their hopes for what CCDS research can change for their affected family member(s). Families used the hashtag #CCDSDay2022 on their social media accounts and research success hopes 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 272 to 350 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, we encourage them to find others online in the established CCDS and GAMT support groups, and we encourage them 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 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 sponsors or special events, which provided funding for 2022:

Contributors

Sponsors:

American Oak & More Solid Wood Furniture
RPK Tools
The Howieson Family
Alliance Bank
Trey & Heidi Wallis Family
Jeremiah Isaacson
AIzChem
Charles D. Nielsen and Family
The Vandenberg Family
King & Co Real Estate Group
Dan & Nicole Kelley Family
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Sangeetha Iyer
Big Sky Oral & Facial Surgery
Servicemaster of Chippewa Valley
Faye & Ed Larson
Buggy Bath Car Wash
The Prescher Family
Everett Stunz
The Lin Family
JoMar Labs
The Bogar Bunch
Heike Law Office
Wolf's Snacks
The Schreiner Family
Fusion Rehab and Wellbeing
Coastal Carriers Truck Lines LLC
The Strum Locker
Ultragenyx
Charles J. Neilson and Family
Poeschel Accounting LLC
Elison Orthodontics
Dale Poeschel & Sara Hansen
Central Lutheran Church
CTL Foods, INC
Coriell Institute for Medical Research
Charlotte's Dad
Marten

Special Events:

2022 Walk for Strength
Holiday Heroes
2022 CCDS Scientific + Patient Symposium
Comedy for a Cure

Holiday Heroes 2022

2022 was the best year of Holiday Heroes thus far, with over $255,00 raised by nearly 800 donors.

Grants Received

EveryLife Rare Giving Tools and Resources Grant

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."

2022 Board of Trustees:

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Board Chair, Trustee

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Director of Advocacy, Trustee

Erin Coller
Director of Communications, Trustee

Celeste Graham
Director of Education, Trustee

Dan Coller
Director of Finance, Trustee

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Randy Allen
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Dr. Andreas Schulze, M.D., Ph.D., FRCPC
Dr. Sylvia Stockler, M.D.
Dr. Sarah Young, Ph.D.
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Dr. Gajja Salomons, Ph. D.
Dr. Matthew Skelton, Ph.D.

ACD 2022 Financial Accounting

Statement of Financials
January to December 2022
Balance

Liabilities & Net Assets

Net Assets, Beginning of Period	$ 564,897
Net Income	$ 95,054
Net Assets*	**$659,951**

*Net Assets Include

Total Assets, End of Period	$ 685,374
Assets With Donor Restrictions	$ 353,386
Assets Without Donor Restrictions	$ 301,565
Total Liabilities	- $ 25,423

Statement of Financials
January 1 through December 31, 2022
Balance

Ordinary Income/Expense

Income

Grants and Contributions	$ 582,806
Contributed Services	$ 18,000
Special Event Revenue	$ 95,234
Investment Income	-$ 4,435
Other	$ 21,089
Total Income:	**$ 712,694**

Expense

Program Services	$ 497,598
General and Administrative	$ 65,712
Fundraising	$ 36,330
Total Expense	**$ 617,640**
Net Income	**$ 95,054**

2022 Audit and Form 990 prepared by: Mullins P.C., Certified Public Accountants