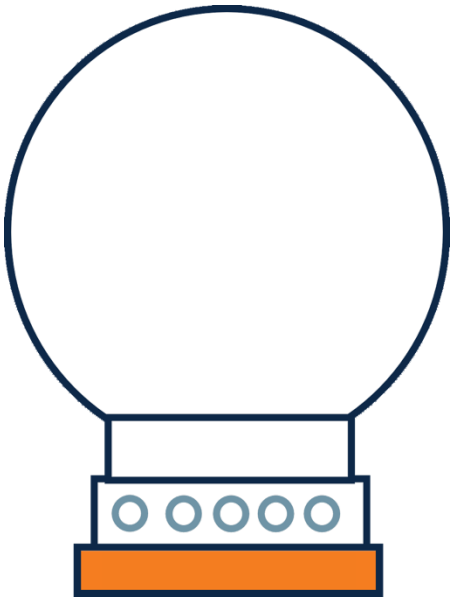
2020 Annual Report

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.

President's Letter

2020 was a unique and challenging year. For much of the world it was a year of struggle, conflict, and loneliness. The CCDS community is uniquely adapted to challenges. In 2020 we came together and advanced research and the ACD organization like no other year!

From the largest Holiday Heroes fundraiser for CCDS research we've ever held, to powerful images of community members around the world walking alone during this year's Walk for Strength, we remained strong and determined in our focus on never stopping in our efforts to improve the lives of those affected by creatine deficiencies and supporting one another.

We canceled our 2020 in-person conference and instead held a virtual conference. We learned through this experience that there is power in bringing together diverse researchers to discuss creatine deficiencies- especially made possible by an easily accessible virtual platform.

Families faced personal struggles and challenges in receiving the care their children needed, but the support group was stronger than ever, and new members continued to find our community and receive the support they needed. New patients continued to receive CCDS diagnoses and their families found comfort in joining our community during this exceptional time of need.

In short, our community proved their strength. I could not be more proud of this community and the organization, ACD, that serves them.

Here's to an even better 2021 and beyond,

Heidi Wallis

President, Association for Creatine Deficiencies

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, 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)

Website and Social Media

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

2020 Social Media Highlights

- Over 10k website visitors
- 1,533 Facebook likes and follows
- 613 Instagram followers
- 248 Twitter followers

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. This year on CCDS Day, families joined together under the theme “What CCDS Research Means to You”. 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.

Advocacy Efforts

Global Genes Annual Meeting: ACD sent representative Faith Ochoa to the Global Genes annual meeting, which is the largest gathering of rare disease stakeholders in the world. This incredible meeting merges patients, caregivers and thought leaders and offers strategic sessions on patient advocacy.

Newborn Screening Efforts: ACD attended virtual newborn screening meetings throughout 2020, speaking on the importance of expanding newborn screening panels, especially to include GAMT Deficiency. ACD representatives also attended EveryLife Foundation for Rare Diseases' Newborn Screening Bootcamp.

Legislation

Newborn Screening Saves Lives Act
HB 1054

Medical and Scientific Research

2020 was a busy year in promoting research efforts for the Association for Creatine Deficiencies.

Research Meetings

2020 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. The conference was attended by 180 people from 17 different countries.

Gene Therapy Consortium: In the winter of 2019, our parent community raised an initial fund of $50,000 to be used towards gene therapy research efforts. With this support in mind, ACD established the CCDS Gene Therapy Consortium in 2020 to foster concentrated efforts into gene therapy for creatine deficiencies.

As a first step, ACD brought together a group of outstanding researchers with expertise in gene therapy and/or creatine deficiencies to form the Gene Therapy Consortium. The consortium includes Dr. Jagdeep Valia, Dr. Andreas Schulze, Dr. Nicola Longo, Dr. Laura Baroncelli, Dr. Matt Stockler, Dr. Gerry Lipshutz, Dr. Olivier Braissant and Dr. Steven Grey. Several of these researchers have been engaged in gene therapy research, in either CTD or GAMT, and are sharing their expertise and knowledge with the entire consortium.

The mission of the consortium is to facilitate the timely sharing of information and development tools among labs that are pursuing gene therapies for creatine deficiencies. By building a collaborative environment and supporting shareable tools through grants, we can shorten the timeline and effort required to find gene therapy solutions for creatine deficiencies. The consortium meets on a quarterly basis as a group to discuss the latest research and provides peer expertise. Sangeetha Iyer, ACD Scientific Advisor, and Laura Trutoiu, ACD Director of Research, facilitate the scientific meetings and follow-up conversations while ensuring that the parent voice is represented.

In February 2020, ACD held the first CCDS gene therapy consortium meeting to discuss progress on gene therapy approaches for CCDS and the outstanding problems faced by the field.

The first meeting shared information on the vector types, i.e. the viral delivery methods that are being developed to generate the gene therapies, and the pros and cons of each method. This fostered a discussion around the outstanding questions in the field:

- What are the levels of creatine that need to be achieved in the brain and elsewhere for global correction?
- How do cells and tissues regulate the intracellular concentrations of creatine?
- What approaches could be utilized to answer these questions?

Understanding these factors will aid in the development of a safe gene therapy for CTD. Given the shared metabolic pathway for creatine and GAMT, such findings could also be extended to GAMT gene therapy approaches.

With the information learned through these conversations, ACD has created the Gene Therapy Advancement (GTA) Award. Each grant has a limit of $10,000 and is used towards capacity building, resource/tool creation, or fulfillment of experiment.

Research Grants

The ACD awarded two Gene Therapy Advancement Awards in 2020:

Q2 Gene Therapy Advancement Award: In June 2020, ACD awarded the first GTA grant to Dr. Laura Baroncelli, Italy, to facilitate capacity building efforts for her lab, specifically focused on the incorporation of intrathecal viral delivery techniques in mice. Intrathecal administration is a route of administration for drugs via an injection into the spinal canal so that it reaches the cerebrospinal fluid (CSF) and is useful for drugs that need to reach the brain. With this award, Dr. Baroncelli's lab will be able to test new viral vectors for expression and subsequent correction of defects in the brain. It's worth noting that learning this technique will require skills from another consortium member, Dr. Steven Grey, who generously volunteered to share his expertise.

Q3 Gene Therapy Advancement Award: In October 2020, ACD awarded the second GTA grant to Dr. Gerry Lipshutz for his ongoing efforts in gene therapy for Guanidinoacetate Methyltransferase (GAMT) deficiency disease. GAMT deficiency is one of the two common cerebral creatine deficiency syndromes arising due to autosomal recessive mutations in the GAMT gene. The current standard for GAMT therapy is life-long and includes oral creatine to replenish cerebral creatine along with supplementation of ornithine and restriction of dietary arginine and protein.

AGAT-Inhibitors Drug Development Research Project: In the final quarter of 2020, ACD was pleased to commit their support to Dr. Nicola Longo in his research into new treatments and therapies for Guanidinoacetate Methyltransferase (GAMT) Deficiency. $25,000 of the funds raised from Holiday Heroes 2020 were dedicated to this research initiative.

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.

2020 Walk for Strength

The 2020 Walk for Strength had 170 walkers from the US, Germany, and the UK. 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. Total funds raised: $18,174.

CCDS Day Campaign

Over 15 families participated in ACD's CCDS Day Campaign, as each family shared what CCDS research means to them. CCDS Day continues to raise awareness and serves as an opportunity to unite the three disorders together for a shared cause.

CCDS Patient Growth

Over the calendar year of 2020, ACD had a 32% growth rate for the entire community. 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

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 ACD's mission.

The following is a list of areas of sources, which provided funding for 2020:

Contributors

Corporate:

Microsoft

Sponsors:

Ultragenyx
TriWest
Alzchem
Edelife
Jo Mar Labs
Jnana Therapeutics
1-800 Contacts

Special Events:

Amazon Smile
Holiday Heroes Outreach
2020 Walk for Strength
Rohan's Research Blend Tea (Friday Afternoon)
Reid's Blend Coffee (Greater Goods)
PatientStrong™

Grants Received

Chan Zuckerberg Initiative (CZI) Rare As One grant

Society for the Study of Inborn Errors of Metabolism (SSIEM) 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."

2020 Board of Directors:

Heidi Wallis
President

Whitnie Strauss
Vice President

Kim Tuminello
Director of Advocacy

Melissa Parker
Director of Finance

Laura Trutoiu
Director of Research

Celeste Graham
Director of Education

2020 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 2020 Financial Accounting

Statement of Financials
January – December 2020
Balance

Assets

Current Assets	$714,166.67

Liabilities & Equity

Equity

Opening Balance Equity	$60,829.25
Retained Earnings	$102,939.05
Net Income	$542,665.07
Total Equity	**$706,433.37**

Total Liability & Equity

As of December 31, 2020	$714,166.67

Statement of Financials
January – December 2020
Profit & Loss

Ordinary Income/Expense

Income

Foundation Grants	$55,922.52
Individual Contributions	$12,477.63
Corporate Contributions	$34,431.16
Product Sales	$1,191.53
Special Event Revenue	$10,682.59
Investment Income	$2,786.86
Program Service Fees	$90.00
Total Income:	**$117,582.29**
Gross Profit	**$117,582.29**

Expense

Personnel	$38,097.03
Program	$732.23
Professional Fees	$30,278.53
Operations	$28,305.34
Total Expense	$95,948.67
Net Operating Income	**$21,633.62**
Other Income	**$521,031.45**
Net Income	**$542,665.07**

2020 Financials prepared by: Melissa Parker, ACD Director of Finance