



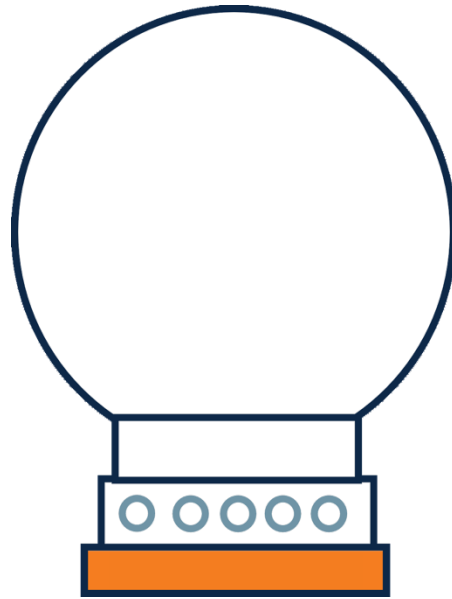
# 2019

## 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

Dear CCDS Stakeholders,

As I review 2019 and all the accomplishments that have taken place over the course of the year, I'm reminded of how far we've come. In the pages that follow, you will see specific and, most importantly, measurable results of twelve more months of hard work and dedication to a cause that impacts us all so much.

The Association for Creatine Deficiencies works for the patients. It is for this reason that we continue to pursue newborn screening, treatments, new research opportunities and partnerships that will one day change the lives of those living with CCDS.

Each year we learn and grow as an organization and find ourselves raising the bar and setting new goals. 2019 has been no different. I am privileged to have completed my term as President with so many wonderful success stories to share. Please join us in celebrating a wonderful year end to 2019 and welcome Heidi Wallis as she steps into her new role as President of the ACD.

Thank you for your continued support.

A handwritten signature in black ink that reads "Whitnie Strauss". The signature is written in a cursive, flowing style.

Whitnie Strauss

# Education and Awareness

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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



## Medical and Scientific Conferences

ACD feels that with a continued presence at strategic medical and scientific conferences we can increase CCDS awareness and promote earlier screening and diagnostics for GAMT, AGAT and CTD patients. Conference and/or speaking engagements for 2019 included:

**ACMG Annual Clinical Genetics Conference:** (3,000+ members of the medical genetics community)

**Drug Information Association (DIA):** Laura Trutoiu, ACD Director of Research, spoke as chair of a panel speaking about collecting better patient experience data. ACD Ambassadors, Celeste and Erin, and ACD Director of Advocacy, Kim Tuminello, were also in attendance.

**Rare New England:** Celeste Graham, ACD Ambassador, was in attendance at the Rare New England Conference in Portland, Maine, on October 26th, 2019.

**Child Neurology Society (CNS):** Whitnie Strauss, ACD President, Andrea Becerra, ACD Marketing and Operations Assistant, along with ACD Ambassadors, Celeste Graham and Regina Bogar, represented the ACD as we exhibited at the 48th Annual Meeting of the Child Neurology Society in Charlotte, NC. (Over 1,100 in attendance.)

**National Society of Genetic Counselors (NSGC):** Salt Lake City, UT, ACD Vice President, Heidi Wallis, and Andrea Becerra, ACD Marketing and Operations Assistant, Andrea Becerra

shared CCDS information with over 2,600 attendees regarding the diagnosis and early intervention that is critical to establishing treatments needed to improve life quality. This conference showcased advancements across the breadth of the genetic counseling profession to provide education and build community.

**World Orphan Drug Congress USA:** The World Orphan Drug Congress is a global gathering of 1,500 leaders in orphan drugs from 50 countries, focusing on the most pressing challenges and opportunities to bring rare disease therapies to patients faster. Laura Trutoiu, ACD Director of Research, discussed the benefits of having three rare diseases under one umbrella.

## Website and Social Media

ACD continues to utilize the website [www.creatineinfo.org](http://www.creatineinfo.org) and our social media channels to increase educational and key messaging.

### 2019 Social Media Highlights



- 1,302 Facebook likes and followers
- Facebook Total Post organic reach: 207,102
- Facebook Total Post Impressions by people who have liked our page: 157,281
- 284 Instagram followers
- 166 Twitter followers

## 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 so important to continue participating in awareness days to join forces 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.

## Other Opportunities

The ACD translated our Understanding CCDS educational brochure in German and Dutch to increase international accessibility to the clinical symptoms that could lead to an earlier diagnosis.



# Advocacy

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The ACD is the only patient advocacy group in the United States that devotes their efforts to changing the lives of people living with CCDS. Along with education and awareness, the ACD is passionate about advocating the rights of individuals with CCDS.

## Newborn Screening

The ACD conducts regular newborn screening committee meetings to discuss policy, engagement opportunities and strategic initiatives. Newborn screening engagements in 2019 have included the following:



**Newborn Screening Bootcamp:** Everylife Foundation & Baby's First Test

**Newborn Screening Lecture with Ton DeGrauw:** ACD Scientific Medical Advisory Member (SMAB)

**Arkansas Department of Health Newborn Screening Advisory Committee Meeting:** Grant Tuminello advocated for newborn screening by telling his personal story.

**Georgia Department of Health Advisory Committee Meeting:** Missy Klor attended the Newborn Screening Advisory as an advocacy representative.

**Georgia Department of Health Advisory Committee:** Unanimously voted to add GAMT to their state newborn screening pending funding.

**House passed Act:** Newborn Screening Saves Lives Reauthorization Act was passed.

## Other Advocacy Efforts

**Coffee with Congressmen:** (San Marcos, CA) Kim Tuminello attended with Congressman Peters.

**NORD Rare Action Network Event & “Share Your Rare”:** Recap Blog Post - Regina

**Tour of Illumina:** Kim Tuminello, ACD Director of Advocacy, and Erin Coller, ACD Ambassador, had the pleasure of touring the Illumina Headquarters to get hands-on experience with their genetic sequencing instruments. These instruments are used for genetic testing purposes including Whole-Genome Sequencing, the most advanced genetic testing option and a comprehensive method for analyzing entire genomes. Genomic information has been instrumental in identifying inherited disorders such as Cerebral Creatine Deficiency Syndromes.

**Mountain State Regional Network:** Heidi Wallis, ACD Vice President, presented on genetic screening for children with developmental delays in Albuquerque, NM.

**Global Genes Annual Meeting:** The ACD sent representatives Kim Tuminello, Melissa Parker, Laura Trutoiu and Andrea Becerra 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.

**Global Genes Rare Foundational Alliance Meeting:** The ACD participated in the Global Genes RARE Alliance Meeting in 2019 to discuss initiatives related to advocacy, training and patient support.

**Nord Rare Summit:** Regina Bogar, ACD Ambassador, was in attendance at National Organization for Rare Disorders (NORD) Rare Summit in Washington, DC, on October 21-22, 2019.

## Rare Disease Partners

As part of our responsibility to advocate for CCDS, the ACD has partnerships with Global Genes, NORD, Child Neurology Foundation, EveryLife Foundation, and ThinkGenetic. These relationships are incredibly important to building an extensive network of Rare Disease advocates who can provide an even greater platform to provide resources, share communication, petition on behalf and represent for our CCDS patients and community.



# Medical and Scientific Research

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2019 was a busy year in promoting research efforts for the Association for Creatine Deficiencies. From organizing and hosting scientific talks to taking the first steps in being listed as co-authors or acknowledged for grant support, the ACD is making a name for itself.

## Research Meetings

**Inborn Cerebral Creatine Deficiency Syndromes Symposium Co-located with SSIEM:** The ACD, efforts led by Heidi Wallis, co-organized and spoke on the patient perspective at a CCDS Symposium in Rotterdam, The Netherlands. This event brought together leading experts in the field to discuss latest advancements and research as it relates to all three creatine deficiencies.

**Society of Inherited Metabolic Disorders (SIMD):** In April 2019, the ACD organized a satellite session at the Society for Inherited Metabolic Disorders annual meeting in Seattle, Washington. A full afternoon of talks on the current treatment guidelines for all three creatine deficiencies took place.

## ClinGen Partnership to Share Genomic Information with ClinVar

ACD is one of six patient organizations participating in an NIH-funded Clinical Genome Resource (ClinGen) group to share de-identified genomic and phenotypic data with ClinVar (a public archive of reports of the relationships among human variations and phenotypes). ClinGen's goal is to make valuable genetic data accessible to the genomics community. ACD community members control their participation via online consent, they provide phenotype information through surveys, then genomic variant information is extracted by the GenomeConnect team from uploaded clinical testing reports. This collaboration demonstrates the value of patients as an important source of genomic data; from the first 100 participants of all six organizations, a total of 81 unique variants were submitted to ClinVar; of these 45.7 (n=37/81) were novel to ClinVar. This project contributed to three papers (references below) that lead author Juliann Savatt presented at different venues

including the largest professional conference for genetic counselors, the Annual Education Conference for the National Society of Genetic Counselors.

1. Savatt, J.M., Azzartiti, D.R, Faucett, W.A., Florin, J., Ledbetter, D.H., Miller, V.R., Palen, E., Rehm, H., Rhode, J., Rogers, L., Talbird, S., Trutoiu, L., Vidal, J.A., Waggoner, C., Riggs, E.R., Martin, C.L. *Expanding Patient Data Sharing: GenomeConnect's Pilot to Engage External Registries in Data Sharing*. Poster presentation. Presented at the 38<sup>th</sup> Annual Education Conference for the National Society of Genetic Counselors, November 6, 2019, Salt Lake City, Utah.
2. Savatt, J.M on behalf of the GenomeConnect team. *ClinGen: Enabling Patient-Driven Data Sharing through GenomeConnect*. (Fall 2019). *American College of Medical Genetics Fall News Magazine*.
3. Savatt, J.M, Azzartiti, D.R, Faucett, W.A., Ledbetter, D.H., Miller, V.R., Palen, E., Rehm, H., Rhode, J., Rogers, L., Talbird, S., Trutoiu, L., Vidal, J.A., Riggs, E.R., Martin, C.L. *ClinGen's Patient Data Sharing Program: Leveraging Data Sharing Experience from GenomeConnect to Broaden Patient Data Sharing Efforts*. Poster Presentation. *Curating the Clinical Genome Meeting*. May 30, 2019. Washington, DC.

## Research Grants

The ACD awarded a Research grant to Dr. Matthew Skelton at the 2018 CCDS Patient + Scientific Symposium. The partial support from ACD enabled Dr Skelton to gather preliminary data and publish several papers (references below) that showcase promising results and potential for gene therapy applicability.

1. Abdulla, Z.I., Pennington, J.L., Gutierrez, A., and Skelton, M.R. (2019) *Creatine transporter knockout mice show increases in serotonin-related proteins and show resilience to learned helplessness*. *Behav. Brain. Res.* PMID: 31542396
2. Abdulla, Z.I., Pahlevani, B., Lundgren, K.H., Pennington, J.L., Udobi, K.C., Seroogy, K.B., and Skelton, M.R. (2019) *Deletion of the creatine transporter in dopaminergic neurons leads to hyperactivity in mice*. *J. Mol. Neurosci*; PMID: 31520365
3. Udobi KC, Delcimmuto N, Kokenge AN, Abdulla ZI, Perna MK, Skelton MR. *Deletion of the creatine transporter gene in neonatal, but not adult, mice lead to cognitive deficits*. *J Inherit Metab Dis.* 2019 Sep; 42(5):966-974;PubMed PMID: 31209903.

## Gene Therapy Consortium and Research

In 2019, the ACD began investigating how to establish a Gene Therapy Consortium. In doing so, the ACD engaged eight world-class scientists, from four countries, that will participate and collaborate to share data and methodology. With the help of our community, the ACD raised an initial sum of \$50,000 to fund collaborative projects in gene therapy. The Consortium will meet quarterly to determine goals and deliverables for 2020.

For more information, visit: [www.creatineinfo.org/research](http://www.creatineinfo.org/research)

# Patient and Family Supportive Services

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The 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.

## 2019 Walk for Strength

The 2019 Walk for Strength grew in 2019 to consist of 21 total teams. 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. The ACD was thrilled to see 366 total participants in 2019, consisting of representation from 2 countries (Germany and the U.S.) and 11 U.S. states!

Total funds raised: \$19,000.

## CCDS Day Campaign

Over 30 families represented in 6 countries participated in ACD's 'We Are CCDS Superheroes' Campaign, as each family shared their child's strengths to raise CCDS awareness on this special day. 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 2019, ACD had a 30% 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 and encourage them to find others online in the established CCDS and GAMT support groups.

# Fundraising

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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 2019:

## Contributors

### Corporate:

United Way  
Microsoft

### Sponsors:

Lumos Pharma  
Jnana Therapeutics  
Ultragenyx  
Alzchem  
Elison Orthodontics  
JoMar Labs  
Admera Health  
TriWest  
Big Sky Ortho Surgery  
1-800-Contacts

### Special Events:

Amazon Smile  
Holiday Heroes Outreach  
Jerry Robinson Ironman  
Beth Robinson Endure for a Cure  
Rohan's Research Blend Tea (Friday Afternoon)  
Reid's Blend Coffee (Greater Goods)

Patient**Strong**™

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

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The ACD was founded in 2012 by parents with children diagnosed with a CCDS. The 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.

The 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."

## 2019 Board of Trustees:

### **Whitnie Strauss**

President, Trustee

### **Melissa Parker**

Director of Finance, Trustee

### **Heidi Wallis**

Vice President, Trustee

### **Laura Trutoiu**

Director of Research, Trustee

### **Kim Tuminello**

Director of Advocacy, Trustee

## 2019 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 2019 Financial Accounting

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## Statement of Financials January – December 2019 Balance

### **Assets**

Current Assets	\$165,370.81
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### **Liabilities & Equity**

#### **Equity**

Opening Balance Equity	\$60,829.25
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Retained Earnings	\$62,761.66
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Net Income	\$40,177.39
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<b>Total Equity</b>	<b>\$163,768.30</b>
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### **Total Liability & Equity**

As of December 31, 2019	\$165,370.81
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Statement of Financials  
January – December 2019  
Profit & Loss

**Ordinary Income/Expense**

**Income**

Foundation Grants	\$3,232.23
Individual Contributions	\$42,885.34
Corporate Contributions	\$73,667.55
Product Sales	\$647.43
Special Event Revenue	\$9,901.85
Investment Income	\$1,379.26
Unrealized Gain/Loss	(\$3,571.98)
Other Income	\$483.60

**Total Income:** **\$128,625.28**

**Gross Profit** **\$128,625.28**

**Expense**

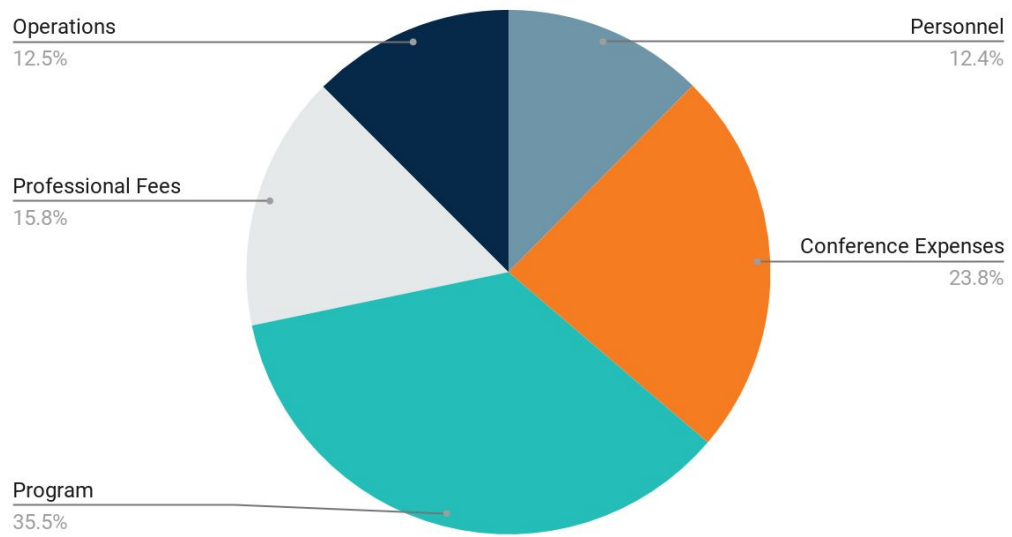
Personnel	\$14,405.58
Conference Expenses	\$27,688.28
Program	\$41,213.23
Professional Fees	\$18,306.98
Operations	\$14,522.10
Total Expense	\$88,447.89

**Net Operating Income** **\$40,177.39**

**Net Income** **\$40,177.39**

## Expense Summary January-December 2019

### 2019 Financial Summary



2019 Financials prepared by: Melissa Parker, ACD Director of Finance