ACD ANNUAL REPORT 2015

ACD
ASSOCIATION FOR CREATINE DEFICIENCIES

Our Mission

The ACD is the only patient advocacy group that is devoted to Cerebral Creatine Deficiency Syndromes (CCDS).

We are committed to meet our goals, which are "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."

The ACD sets these goals to bring the CCDS Community together as one strong voice, to be united in our efforts, to urge allied medical professions to test, and to treat and promote scientific research.

It is through our mission that our vision for the future is clear: to eliminate the challenges of Cerebral Creatine Deficiency Syndromes.

It is with our integrity, community and hopes that we are "building strength and changing lives."


IF YOU WANT
TO GO FAST,
GO ALONE.
IF YOU WANT
TO GO FAR,
GO TOGETHER.
UNKNOWN AUTHOR

President's Letter

Dear Friends,

In 2015, the ACD saw a deeper awareness of CCDS. Our advocacy efforts in our conference attendance at the American College of Medical Genetics and Genomics (ACMG) and the Child Neurology Society (CNS) proved valuable as more and more attendees showed an understanding of AGAT, GAMT and CTD and a commitment to screen for these disorders. The ACD plans to expand these education and awareness opportunities in 2016 to include Autism and Developmental Pediatric audiences.

Newborn screening has been a large focus and will continue to be a priority in the coming year. The state of Utah was the first to pass newborn screening for GAMT in 2015. As this annual report goes to press, I am pleased to announce that GAMT has been nominated for the RUSP (Recommended Uniform Screening Panel) and will be considered by the Advisory Committee on Heritable Disorders in Newborns and Children in Rockdale, Maryland. The ACD will be in attendance and speak on behalf of GAMT along with several CCDS family advocates as it pertains to the importance of adding GAMT to the RUSP. This attendance will be an exciting time and of utmost importance; being accepted could mean that every baby born would be screened for GAMT across the U.S.

And as a related development for newborn screening research, the Mayo Clinic Newborn Bloodspot Project, which is researching technology to screen for CTD, has reached out to us to help in their efforts to collect patient applications. All these ventures are in alignment with our mission.

AGAT, GAMT, and CTD are considered rare and neglected diseases. Our collaboration with the National Organization for Rare Disease (NORD) and Global Genes has been essential. Coming together in support of rare diseases is important as institutions like the FDA are taking notice. In 2015, the FDA approved twenty-one orphan drugs for rare diseases. Orphan drugs are developed to treat rare disorders with a small patient population. These drugs would likely not be advanced without the government's help because of the high cost of making them and a small market population to recover those costs. Collaboration with other nonprofits gives us a larger voice in making reform. We can't continue our mission without the support of our patient community.

This past 2015 fundraising campaigns succeeded our goal. From small donations, in-kind donations, underwriting and corporate giving, these gifts helped us raise over $20,000. These funds will allow us to operate and champion for our shared mission to eliminate the challenges of CCDS through education, advocacy and research.

The ACD Board of Trustees meets monthly to review on-going strategic goals and their progress. As the momentum continues to grow, we will continue to expand our Board of Trustees with an emphasis on bringing together leaders in business and health science.

We will also be building our Scientific Medical Advisory Board to include the best in the CCDS field, to help us advance our efforts.

On behalf of the ACD Board of Trustees, I am honored to present the ACD's Annual Report for fiscal year 2015. As you will see in these pages, we have had a very successful year. As CCDS needs grow, it will require us to be stronger, work harder, and stretch ourselves even further. Based on our progress this past year, I am certain we will succeed.

Thank you for your continued support.

Kim Tuminello

The Importance of Cerebral Creatine Deficiency Syndromes (CCDS)

Let us take a moment to understand this disease. CCDS are inborn errors of metabolism, which interrupt the formation or transportation of creatine. Creatine is necessary to increase adenosine triphosphate (ATP) which provides energy to all cells in the body. Creatine is essential to sustain the high energy levels needed for muscle and brain development.

There are three Cerebral Creatine Deficiency Syndromes with complicated medical names for some to remember. So, we refer to them by acronyms.

Arginine: Glycine Amidinotransferase Deficiency or AGAT

AGAT is involved in the first step of creatine formation. Mutations found in the AGAT gene impair the body's production of creatine. There are fewer than twenty AGAT cases reported, to date.

Guanidinoacetate Methyltransferase Deficiency or GAMT

GAMT is a mutation in the gene that makes the enzyme that creates creatine, resulting in a shortage of creatine. GAMT is the most severe of the three CCDS due to an elevation of guanidinoacetate (which is neurotoxic) in addition to creatine deficiency, and there are more than 100 cases reported in medical literature.

Creatine Transporter Defect or CTD

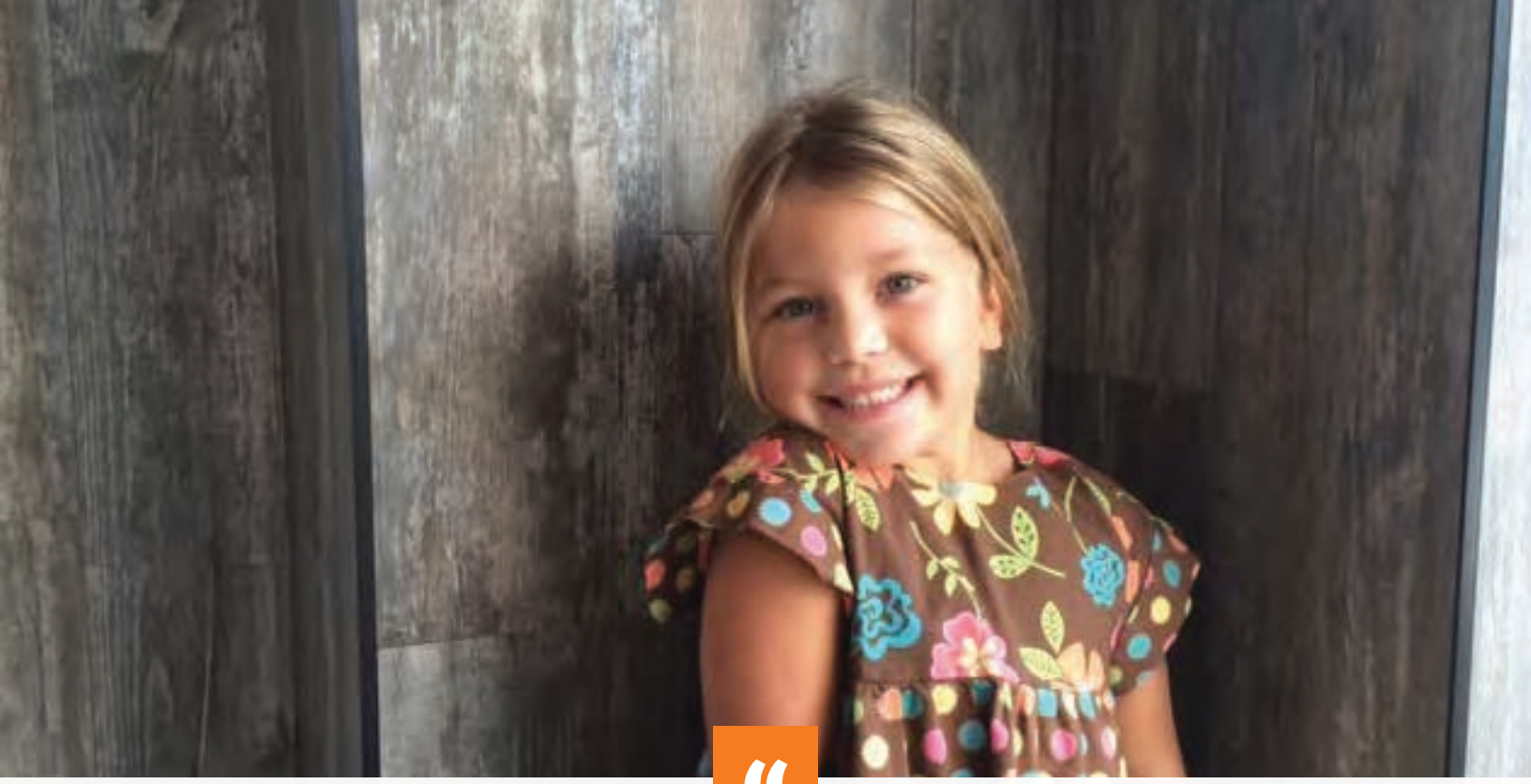
CTD is a mutation in the creatine transporter gene. This mutation results in a blockage in the transportation of creatine to the brain and muscles. CTD is the most common CCDS. It is estimated to account for about 2% of all unexplained X-linked intellectual disabilities.

Symptom(s)

The most common symptom of CCDS is a global delay. Speech delay may be particularly severe, and intellectual disability of variable severity is typically present in older children and adults. Other additional symptoms may include seizure disorders, muscle weakness, autistic-like behaviors, movement disorders, gastrointestinal problems and failure to thrive.

Treatment(s)

Treatment with oral supplementation is available and effective if initiated early for AGAT and GAMT patients. To date, this type of therapy has not shown to improve outcomes in individuals with CTD.



A HERO IS AN ORDINARY INDIVIDUAL WHO FINDS THE STRENGTH TO PERSEVERE AND ENDURE IN SPITE OF OVERWHELMING OBSTACLES.

CHRISTOPHER REEVE

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 has partnered with the National Organization for Rare Disease. This collaboration has helped to strengthen the ACD capacity to serve our patient population, to increase CCDS knowledge within the medical community and, in general, the public at large.

Medical and Scientific Conferences

With this as a major focus, the ACD has been active and instrumental in urging the medical professionals to seek out and test patients for CCDS. By attending and exhibiting at conferences, such as the Child Neurology Society and the American College of Medical Genetics and Genomics, we are helping to advance CCDS knowledge and training.

Autism Speaks

2015 is ACD's first year of participation at the Los Angeles and Orange County Autism Speaks annual walk-a-thons to help raise CCDS awareness within the family and public sector.

Website and Social Media

Within this scope, the ACD website is continually updated with current research summaries, patient resources, up to date facts related to CCDS, our Insider eNews, opportunities to participate in non-drug research programs and family supportive stories, plus much more. We have increased our social media presence on Facebook, Twitter, and Instagram to keep our message and educational interest visible.

Online Video Publication

Recently, the ACD produced an awareness video called “A Short Story About Creatine Deficiencies” that has been distributed worldwide to raise disease awareness for a wider audience. This video has been used not only as a public teaching tool but will be instrumental in our crowd-funding platform slated for the new year.

FDA Patient Network

The ACD is pleased to announce the board’s involvement with the FDA Patient Network (PN) for inborn errors of metabolism. The FDA Patient Network is a comprehensive

program that works to sustain communications with patients and their community. The PN also helps educate patients, patient advocacy groups, and health care professionals about medical product regulations.

Other Opportunities

The ACD has published a detailed, clinical diagnostic booklet along with a family information brochure to aid in our educational efforts.

> We were told there was not much known about the disorder and didn't know if he would progress. They mentioned intellectual impairment, attention deficit, speech and language impairment and seizures. We walked out of that appointment shocked, overwhelmed, and scared about the future.
>
> Mother of 5 year old diagnosed with CTD

Advocacy

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 aims for CCDS to be placed on newborn blood screens nationwide. Newborn screening identifies conditions that can affect a child's long-term health or survival. CCDS early detection, diagnosis, and disease-specific treatment can prevent disabilities and will enable a child to reach their fullest potential.

We are actively pursuing this initiative. Newborn screening has not been an easy challenge, but 2015 has paved the way for a favorable future outcome for GAMT newborns. With a current pilot study in Utah, we are hopeful that catching a positive GAMT patient from a newborn bloodspot will prove the effectiveness of GAMT newborn screening and will encourage a nationwide following.

We believe with our involvement within the CCDS medical community and our advocacy efforts from attendance and exhibit booths at both the Child Neurology Society and the American College of Medical Genetics and Genomics has helped us earn the respect and recognition to support and promote newborn screening.

Rare Disease Partners

As part of our responsibility to advocate for CCDS, the ACD has partnered with Global Genes, one of the leading rare disease patient advocacy organizations in the world. This collaboration has been effective in our training, communication, and growth for the future representation of CCDS patients.

> "Everyday it seemed there was another problem or worsening of her condition, watching her decline to the worst point.
>
> Mother of 6 year old diagnosed with GAMT

"

ONCE YOU CHOOSE HOPE,
ANYTHING'S POSSIBLE.

CHRISTOPHER REEVE

Medical and Scientific Research

The ACD assists approved non-drug research studies for CCDS by promoting volunteer participation.

Patient Registry

This year the ACD is excited to announce our partnership with Patient Crossroads Connect to house our Patient Registry database.

One of the most important aspects of research is a patient registry. The ACD's Patient Registry collects critical information to understand the history and progression of CCDS, to make it easier for researchers to study, and for the patient and family to learn about treatments, and for the ACD to speak on behalf of the CCDS community.

Patient Crossroads Connect collects and safeguards the ACD's data to protect participants' privacy.

With our patient registry data, the ACD has been actively reaching out to the National Institute of Health (NIH), the Center for Disease Control (CDC) and establishing relationships with pharmaceuticals to promote research.

In ending the year 2015, we have been leading the way to secure more interest in CCDS studies, with the first established CCDS patient registry.

> "Beyond the fear, there was the anger. Why us? Why did other people have child after child without any issues? What had we done?
>
> Mother of 6 year old and 14 month old diagnosed with GAMT

Patient and Family Supportive Services

The ACD's website is an essential platform for CCDS information. Our patient and family supportive services can be obtained by visiting our site www.creatineinfo.org. For 2015, we have brought together areas of interest for supportive services. We listen to the needs of our community and take every means to produce helpful and beneficial information for daily use.

PatientStrong™

The ACD is excited to launch our PatientStrong ™ Grant Program. This grant program was established to assist patients diagnosed with a CCDS, by providing funds to help assist with medical expenses. Grants are awarded bi-annually, amounting to $500 each. ACD's PatientStrong ™ Outreach Fundraiser helps support this anticipated award.

Ask The Expert

This year our Scientific Medical Advisory Committee is engaged in the patient participation of CCDS related questions. Our "Ask The Expert" series, featured in our quarterly newsletter, helps to answer questions our community members may have.

Patient Resource Guide

Patient resources are a valuable channel of information that can assist in one's care. With many hours of searching for the best information and resources for CCDS patients and families, we have developed our first ACD Patient Resource Guide for use to our CCDS community. This information is to be used as a tool, to aid in supportive services that may be reviewed, and to complement patient's knowledge and programs. We promote these resources on our website for easier accessibility. We will be annually updating our guide for up-to-date information to pass along to patients and families.

CCDS Family Journey

The ACD has asked the CCDS community to submit their personal stories, to share with others that may be dealing with similar issues. We then post them to our website. These narratives have been received so well, ACD will have a full online blog in the new year to collect all stories submitted. We are honored to be entrusted with such personal stories, and we are grateful to be able to post them.

DON’T CRY WHEN THE SUN IS GONE, BECAUSE THE TEARS WON’T LET YOU SEE THE STARS

VIOLETA PARRA

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 2015:

Contributors

Corporate:

United Way
Microsoft
Mode-ology
Smith-Cooper International

Special Events:

Amazon Smile
Holiday Hero's Outreach
Lobster-Fest Dinner and Social Gala
PatientStrong™

Matching Gift Funds:

Enterprise Rent A Car

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

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.”

2015 Board of Trustees:

Kim Tuminello
President, Trustee

Whitnie Strauss
Vice President, Trustee

Linda Cooper
Administrative Director, Trustee

Missy Klor
Financial Director, Trustee

Melissa Parker
Trustee

Heidi Wallis
Trustee

2015 ACD Scientific Medical Advisory Board:

Dr. Nicola Longo, M.D., Ph.D.
Chief, Division of Medical Genetics
University of Utah Health Care
Pediatrics Medical Genetics

Dr. Bruce Barshop, M.D., Ph.D.
Metabolic and Mitochondrial Medicine
Pediatrics, Rady Children’s Specialty
University of California San Dieg

Dr. Sarah Young, Ph.D.
Assistant Professor
Department of Pediatrics - Medical Genetics
Duke University School of Medicine
North Carolina

Dr. Denise Morita, M.D.
Pediatric Neurologist
Granger Medical Clinic
Riverton, Utah

ACD 2015 Financial Accounting

Statement of Financials
January – December 2015
Balance

Assets

Current Assets	$50,177.88

Liabilities & Equity

Equity

Opening Balance Equity	$23,694.12
Net Income	$26,483.76
Total Equity	**$50,177.88**

Total Liability & Equity

As of December 31, 2015	$50,177.88

Statement of Financials
January – December 2015
Profit & Loss

Ordinary Income/Expense

Income

Donations	$49,160.15
Total Income:	**$49,160.15**

Expense

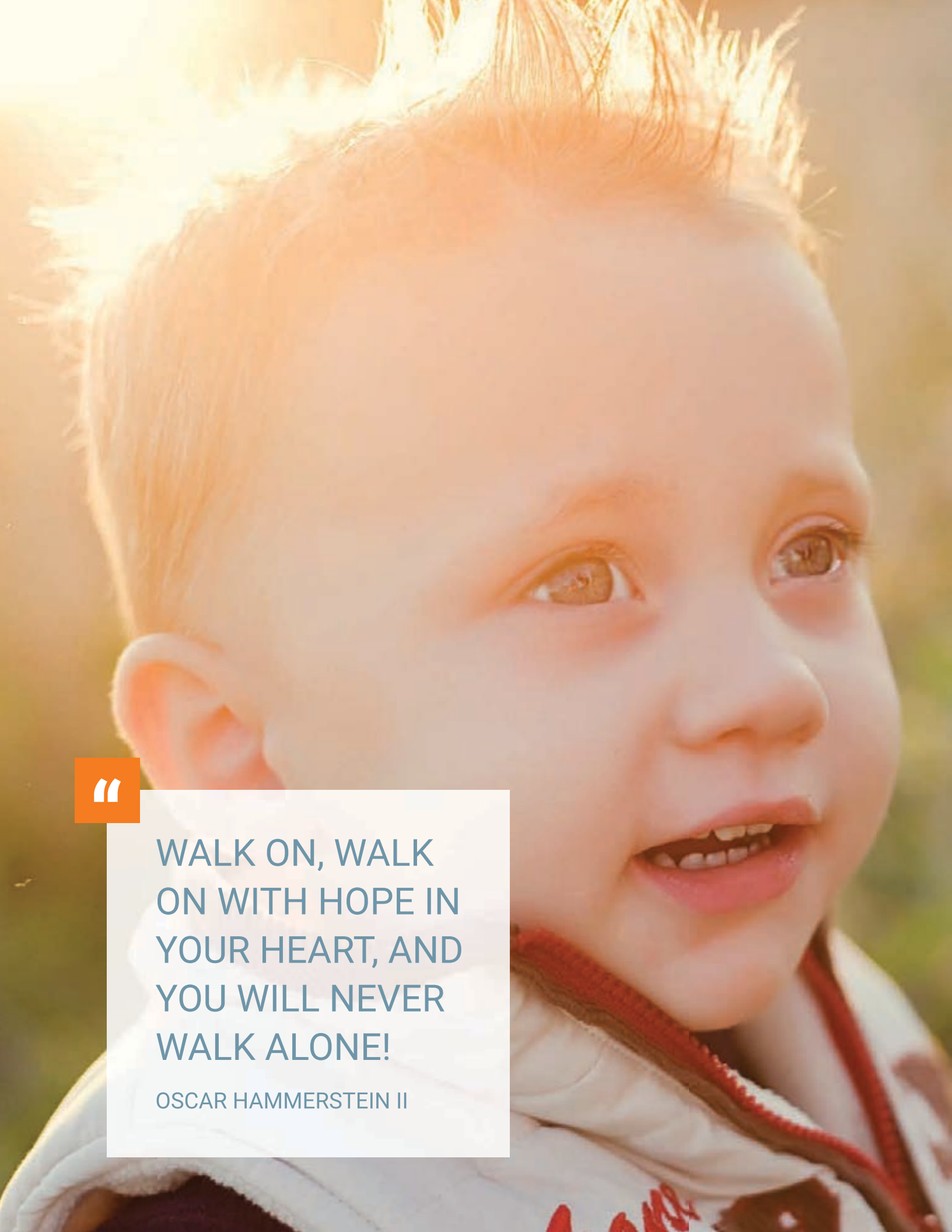
Accounting & Tax	$856.00
Advertising	$2,099.50

Conference(s)	$7,659.19
Contract Services	
Legal Fees	$925.00
Dues & Subscript	$50.00
Fees & Charges	$408.81
Fundraising	$3,179.30
Insurance	$79.00
License & Permits	$45.00
Office Equipment & Supplies	$814.74
Postage & Delivery	$326.45
Travel & Meetings	
Travel	$2,475.90
Web & Social Management	$3,757.50
Total Expense	$22,676.39
Net Ordinary Income	**$26,483.76**
Net Income	**$26,483.76**

Independent Monthly Data: Bookworks,
Costa Mesa, California

Independent Annual Accounting: Titanium Financials,
Irvine, California

Expense Summary
January-December 2015



WALK ON, WALK ON WITH HOPE IN YOUR HEART, AND YOU WILL NEVER WALK ALONE!

OSCAR HAMMERSTEIN II