Rare Disease Day 2017 was a tremendous success for the ACDA. Together with The David Ashwell Foundation, we raised over $5,000! Our sincerest thanks to the families, friends, colleagues and classmates of the ACDA and The David Ashwell Foundation. Please read all the details on page 4!

Regards, Eliza Rista, President
RESEARCH NEWS

NORD GRANT UPDATES:

2016 NORD Grant:

The previously announced 2016 NORD grant (see Issues #55 and #57 of ACDA Notes) in the amount of $50,000 for ACD research was recently awarded to Dr. Przemyslaw Szafranski, PhD at Baylor College of Medicine for the study entitled, "Modeling ACDMPV therapies by targeting negative regulators of FOXF1 and genes outside the SHH pathway."

2017 NORD Grant:

It is the great pleasure of the ACDA and The David Ashwell Foundation to announce a $50,000 grant will also be issued in 2017 for ACDMPV research. NORD announced the Request for Proposals (RFPs) on April 26, 2017 (see the box to the right for additional RFP information).

As background, nine grants for ACDMPV research have been issued through NORD since 2005, including 2005, 2008, 2012 (two grants), 2014 (two grants), 2015, 2016 and 2017. Please visit our website HERE to read a full history of NORD grants for ACDMPV research.

None of this would be possible without the hard work, contributions and fundraising efforts of families and friends affected by ACDMPV. We are deeply grateful for the support as we continue to work towards ending this disease.

NORD Request for Proposals (“RFP”) – 2017 Research Grant:

NORD, with funding from The David Ashwell Foundation and the Alveolar Capillary Dysplasia Association (ACDA), is accepting applications for a total of $50,000, for scientific research studies and/or clinical research studies related to Alveolar Capillary Dysplasia/misalignment of the pulmonary veins (ACDMPV). NORD encourages all U.S. and international researchers interested in studying ACDMPV to consider applying for 2017 funding. The Abstract Submission Deadline is June 23, 2017. See the full RFP and abstract template HERE.

Human Mutation (March 2017):

Our thanks to Dr. Simon Ashwell, ACDA father to David (March 4, 2011 – March 19, 2011), for the following summary of a journal article about ACDMPV published in Human Mutation in March 2017:

Casanova, Monteagugo-Sanchez & Guerineau et al: Maternal mutations of FOXF1 caused ACD despite not being imprinted

This paper from a team at Barcelona University, Spain focuses on imprinting in ACDMPV. We define paternal imprinting in the ACDA genetics guide as ‘the inactivation of a copy of a gene inherited from the father of an infant so that the gene is silent and not expressed. This leaves only the gene copy inherited from the infant’s mother active and expressed.’

We know that around 90% of mutations and 95% of deletions of FOXF1 are on the chromosome the infant inherited from...
his/her mother. It has thus been presumed that FOXF1 is subject to paternal imprinting (of course the majority of FOXF1 abnormalities arise de novo in the process of egg formation and are thus not actually inherited from the mother). However, there have been some findings that have been inconsistent with imprinting. Two prior cases of ACDMPV (both a familial case and a de novo case) have been described which were caused by FOXF1 mutations inherited from the infants’ fathers. Additionally 4 children have been identified who do not have ACDMPV but in whom both copies of chromosome 16 (in which FOXF1 is found) have been inherited from their fathers. If FOXF1 is subject to paternal imprinting this should have resulted in ACDMPV but this was not the case.

The authors of this paper thus investigate imprinting by studying 3 infants with ACDMPV, two identical twins and a separate infant. They found all 3 infants to have FOXF1 mutations that were derived from their mothers. However they then analysed the expression of FOXF1 in a variety of adult and foetal tissues and found that the genes derived from both mother and father were expressed in these tissues. They went on to study FOXF1 methylation. This is the process in which methyl groups are added to genes in order to switch them off. It is a key feature of imprinting. They found no methylation of FOXF1.

The authors conclude that the clinical and genetic data to date do not support the long-held view that FOXF1 is subject to paternal imprinting. They suggest an alternative, currently unknown mechanism is responsible for the maternal inheritance pattern of FOXF1 gene abnormalities.

AWARENESS NEWS

Newspaper Articles (Aurora Splawn and Braylee Speed):

Please read the following two newspaper articles published about two of our ACDA families in connection with Rare Disease Day:

- **In memory of Aurora Sophia Mercedes Splawn** (July 23, 2016 – August 6, 2016)

  Rare Disease Day: A Traverse City Family Shares Their Story of Loss

  9&10 News, February 28, 2017

  Traverse City, Michigan, USA

- **In memory of Braylee Graylyn Speed** (February 13, 2014 – February 26, 2014)

  Remembering Braylee: Family draws attention to rare diseases, support

  The Daily Times, February 26, 2017

  Blount County, Tennessee, USA
Update on Rare Disease Day (February 28, 2017):

We are pleased to announce $2,900 was raised by the ACDA and an additional £1,741 was raised by our sister UK registered charity, The David Ashwell Foundation, for a grand total of $5,016 in connection with Rare Disease Day on February 28, 2017!! Our sincerest thanks to the families, friends, colleagues and classmates of the ACDA and The David Ashwell Foundation for raising such an incredible amount for ACD research!

Please click HERE to check out our ACDA families and friends wearing the ACDA logo and #JeansForGenes on Rare Disease Day!

In the U.S., we would like to give our very special thanks to:

- Moore & Van Allen PLLC (Charlotte, NC)
- Family Design Resources (Harrisburg, PA)
- Little Treasures Learning Center (Grove City, PA)
- The Smihtown Library (Nesconset, NY)
- East Quogue Union Free School District (East Quogue, NY)
- Suffolk County District Attorney, Economic Crime Bureau (Hauppauge, NY)
- Suffolk County Attorney’s Office (Hauppauge, NY)
- Suffolk County Office for the Aging (Hauppauge, NY)
- Children’s Choice Childcare, Inc. (Hauppauge, NY)

In the UK, we would like to give our very special thanks to:

- Brighton & Hove Albion FC
- Moulsoomb primary school
- Lewes priory school
- Durham University (various schools & central services)
- Newcastle University Institute of Health & Society
- Skerne Medical group
- Runway East
- Kirby Great Broughton school
- Yarm primary
- Unforgettable™

We would like to thank all of our individual donors and anyone who wore #JeansForGenes or sent pictures wearing the ACDA logo! Thank you for your outstanding support of our ACDA families.

The ACDA also created flyers for use at workplaces and schools encouraging colleagues and classmates to donate $5 or £1 to wear jeans to work or school on February 28 for Rare Disease Day.
Open letter to our social media followers posted on Rare Disease Day 2017:

What does it mean to be rare? In the case of ACDMPV, it means you are one of only a few hundred children worldwide formally diagnosed with ACDMPV since first identified in 1948. By contrast, the U.S. National Institutes of Health (NIH) considers a disease rare if it has a prevalence of fewer than 200,000 affected individuals in the United States. Taking a moment to reflect on those numbers, you realize how rare ACDMPV is even within the subset of rare diseases. And yet, this very rare disease often feels a little less rare due to the compassion, generosity and support of our ACDA families and friends.

On this Rare Disease Day 2017, we are stopping to thank our ACDA families and friends for listening, learning, sharing and always remembering our children who are no longer with us or faced uncertain futures due to this very rare disease. We thank everyone who has ever ‘Liked’ our Facebook page, wore #JeansforGenes, donated funds for ACDMPV research, dressed in a shirt with the ACDA logo, organized a fundraiser, used the designated hashtags #RareDiseaseDay #WRDD2017 or shared an ACDA link. We also thank anyone who has ever sent a hug or love on those difficult days, spoken our child’s name on an ongoing basis, held our hand through an anxious rainbow pregnancy or any of the other innumerable ways you have shown your support. Our list could go on forever in so many ways.

If there is one glimmer of light in the reason that unfortunately brought all of us together, it is that the ACDA community itself is also rare. Not just because of the statistical numbers, but because our ACDA families and friends are unusually remarkable.

Lights for ACDMPV (Johnny Rista):

The Wells Fargo’s Duke Energy Center in Charlotte, North Carolina, USA was lit on February 20, 2017 in red, purple and blue to represent the colors of the ACDA logo to bring awareness to ACDMPV (tall building on right pictured below). The light campaign was done in memory of Johnny Rista's birthday. Thank you for the support @WFLightsCLT!

Lung Transplant Update (Luca Palmisano):

We wrote about Luca Palmisano in Issues #57 and #58 of ACDA Notes and now he is making the news! Luca was recently the lead story on FOX Chasing News (FOX29 Philly, FOX5NY and My9NJ). Watch the video HERE. Luca was also featured in two of his local newspapers! Click HERE and HERE to read the touching stories in The Trentonian and MercerSpace.

In November 2016, Luca received a lung transplant at 4.5 months old at Children's Hospital of Philadelphia in the USA. You can follow Luca’s Lung Transplant Story HERE. #teamluca
FUNDRAISING NEWS

Donations:

To make a secure tax deductible donation to the ACDA or the NORD Research Fund (ACD), please visit our website for full instructions.

[acdassociation.org/donate](http://acdassociation.org/donate)

The ACDA is a 501(c)(3) non-profit, tax-exempt organization as designated by the Internal Revenue Code of the United States.

Balance of ACDA account:

The balance of the ACDA bank accounts as of May 16, 2017 is $xxx.

Update from AmazonSmile:

The ACDA was issued a $xxx donation from the AmazonSmile Foundation as a result of AmazonSmile program activity between October 1 and December 31, 2016. To designate the ACDA as your charity, please follow the link below so that all of your eligible shopping will benefit the ACDA: [http://smile.amazon.com/ch/46-2915711](http://smile.amazon.com/ch/46-2915711)

Update from Spreadshirt:

The accrued commission payment from Spreadshirt between January 23, 2017 and May 3, 2017 is $xxx. Items with the ACDA logo are available for purchase in our Spreadshirt store [HERE](#).

Update from NORD (2017 Grant):

The NORD balance of the Research Fund (ACD) is $xxx as of May 10, 2017.

The ACDA and The David Ashwell Foundation are pleased to announce the above balance exceeds the required minimum amount ($35,000) for the issuance of a 2017 grant! Please see page 2 for detailed information about the 2017 NORD grant.

Let’s start work on a 2018 grant!

Donations Received:

Thank you to the following families and friends that have made donations to the ACDA since the last ACDA Notes:

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[acdassociation.org/donate](http://acdassociation.org/donate)

The ACD is a 501(c)(3) non-profit, tax-exempt organization as designated by the Internal Revenue Code of the United States.
*We are sorry we do not know the child for whom the honorary contribution was made. Please contact us to let us know.

**In honor of Merrideth Lilly Kateridge:**

The ACDA extends its thanks to Alexis Kateridge, ACDA mom to Merrideth Lilly Kateridge (March 9, 2017 – April 5, 2017). Alexis is in the process of organizing various fundraisers for ACD research in honor of her daughter, Merrideth. Please click [HERE](#) to read more.
FJNortheasters, patch auction, hockey game (Fallon Rilling):

Once again, it’s fundraising season for the Toyota off road club, the FJNortheasters.

Each year, this awesome family of “Off Road” enthusiasts gets together for a huge charity off-roading event. April 28-30th was this year’s dates, and the event didn't disappoint. The club raised a significant amount of money, and after all expenses were paid, the proceeds were split between the Make A Wish Foundation and the ACDA. A total of $2,444 was donated to the ACDA in honor of Fallon Jade Rilling. A huge thanks to the club’s president, Richard LaRusso, and the rest of the staff, sponsors and members for making such a large impact in our lives.

May was a huge donation month in honor of Fallon. Additionally, one of the members from the FJ Northeasters club, Greg Muzila of GZiladesigns, held a "patch auction" where he and others donated rare and popular velcro back patches (yes, people collect patches) to be bid on. Over $1,000 was raised with the patch auction, along with the sale of some special "444" patches and decals designed by club members Holly Pulis and Kenny Correa. The 444 patches were produced by Mark of Amaesing Decals.

To continue the fundraising efforts, Fallon’s dad, Bob Rilling, played in a charity ice hockey game on Saturday, May 6th, between the Suffolk County Police Department versus the Suffolk County Corrections Department. Not only did Bob’s team (SCPD) select the ACDA as their charity of choice, but in a heartwarming moment before face-off, the opposing team announced they would be donating their portion, as well, to the ACDA in honor of Fallon. The event, after expenses, raised over $1,000.

We are so proud of Fallon and how much her short life has affected those around her, who continue to support her and the ACDA. Here’s to saving more lives and helping to find a cure.
Rotterdam Marathon, Stichting ACD, Book and Bingo (Myla Lourens):

Mathijs Lourens, ACDA father to Myla (November 20, 2015 - December 17, 2015), ran a full marathon in Rotterdam on April 9, 2017 (you may remember Issue #58 of ACDA Notes featuring Mathijs' half-marathon in Amsterdam in October 2016)! In his own words about the Rotterdam race, “Well today was another very special day. I have been working hard this last year to run my first marathon for Myla, for ACD. And today I did it. It was hot and it was hard but Myla, my family and my friends gave me superpower once again. It feels great to get so much support and it keeps me going. I will continue running for ACD, for the ACDA and my own foundation Stichting ACD. I even did a short interview on a Dutch local television about Myla and ACD because I walked with Myla’s picture to the finish…Oh one more thing. We already raised about €4000 for ACD in the last two months!!”

In addition to his work on the marathons, Mathijs has written a book and started a Dutch ACD foundation called “Stichting ACD”. Please visit the website at the following address (https://stichtingacd.nl) to learn more and you are also invited to view the “Angel Gallery” for a touching image display featuring many of our ACD babies.

If you would like to order a copy of Mathijs’ book describing his experience of being Myla’s father, the book is for sale and the proceeds benefit his Dutch foundation.

The beautiful t-shirts Mathijs and his family and friends wore for the Rotterdam marathon were designed by fellow Dutch parent, Roelina Jut, ACDA mom to Jasmijn (February 8, 2013 - February 23, 2013).

Finally, Mathijs and Jessica Anderson (Myla’s mom) organized and hosted a neighborhood Bingo fundraiser in March 2017. Gifts were donated by shops and restaurants and the event raised over €600!
NEW ACDA BOARD:

Please help us in welcoming our updated Board of Directors as part of a planned transition effective as of February 28, 2017. During a Special ACDA Board of Directors meeting in February 2017, the following individuals were appointed to the Board of Directors of the ACDA for the remainder of the 2016-2017 term:

- President – Eliza Rista
- Secretary – Renee Murray
- Treasurer – John Rista

The ACDA President, Eliza Rista, has previously served as ACDA Secretary since August 2015. The new ACDA Secretary, Renee Murray, has served as a member of the ACDA Fundraising Committee since January 2016.

Below are the 2017 Communication Committee and Fundraising Committee members:

**Communications Committee:**
- McCayla Butler
- Emily Eschweiler
- Diana Woida Locke
- Candice McCasland
- Eliza Rista

**Fundraising Committee:**
- Renee Murray
- Kristen Rilling
- John Rista
- Stephanie Smith

Please check our website for a full listing of Board and Committee members and let us know if you would like to get involved!

[http://acdassociation.org/board-members](http://acdassociation.org/board-members)

Thank you to Steve and Donna Hanson:

As part of the leadership transition, the ACDA would like to take a moment to sincerely thank Steve and Donna Hanson for their many years of leadership, commitment and service to the ACDA.

The Hansons are parents to Eric Stephen Hanson (June 7, 1997 – June 17, 1997) and led the ACDA since 2002. Their contributions to the ACDA have impacted every facet of this organization, including database management, newsletters, research coordination, awareness and outreach, fundraising and culminated in the ACDA obtaining 501(c)(3) nonprofit status in 2014. **The ACDA is forever grateful to the path carved by the Hansons in memory of their son, Eric.**

The above commemorative statue was presented to the Hansons in February 2017.
Welcome to New Families

A sad but warm welcome to the following newly registered family:

- [Name]
- [Name]
- [Name]
Safe Arrivals!

Congratulations on the birth of the following little siblings in our ACDA registered families:

- [List of siblings]

CONNECT WITH US

Facebook:
- Official ACDA Public Page
- Parent Group (private)
- Family Group (private)

Read about the private groups with information on how to join: http://acdassociation.org/support-groups/

Twitter:
- Follow us @acdassociation

Website:
- acdassociation.org

Email:
- President@acdassociation.org (Eliza Rista)
- Secretary@acdassociation.org (Renee Murray)
- Treasurer@acdassociation.org (John Rista)

A note from the President: We absolutely want to hear from you as to how we can best meet your needs with respect to information about ACDMPV and also grief support. We are here to help in any way we can. Please know we always want to hear your ideas and we love community involvement on any level. Please never hesitate to contact me at President@acdassociation.org.

Regards, Eliza Rista, mom to Johnny (February 20, 2013 – March 4, 2013)