Rare Disease Day 2018 was a tremendous success for the ACDA. Together with The David Ashwell Foundation, we raised over $5,600! 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 3!

Regards, Eliza Rista, President
RESEARCH NEWS

2017 NORD Grant Update:

Below please see a write-up of the progress made with respect to the 2017 NORD grant (see Issues #59 and #58 of ACDA Notes), as written by grant recipient, Arun Pradhan, PhD, at Cincinnati Children's Hospital Medical Center in Cincinnati, Ohio, USA for the study entitled, "Development of FOXF1-activating small molecule compound for the treatment of Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins (ACDMPV)."

"In the recent study, we have focused on generation of clinically relevant mouse model of ACDMPV (S52F-Foxf1). Even though ACDMPV is linked to the mutations in the FOXF1 gene the molecular mechanism of pathogenesis of ACDMPV is poorly understood. We have recently dissected the molecular mechanism of S52F-Foxf1 knock-in mice generated in our laboratory by using CRISPR/Cas9 technology. The manuscript depicting the molecular mechanism of pathogenesis due to S52F-Foxf1 mutation is under review. In this report we have shown that the S52F FOXF1 mutation disrupted STAT3-FOXF1 protein-protein interactions and inhibited transcription of Stat3, a critical transcriptional regulator of pulmonary endothelial proliferation and angiogenesis. Also, the S52F-Foxf1 knock-in mice fully recapitulated histopathological findings in ACDMPV infants, including lung hypoplasia, alveolar capillary dysplasia, misalignment of pulmonary veins, increased mortality and gall bladder agenesis. Furthermore, we have developed a novel formulation of cationic nanoparticles, which is highly efficient in delivering gene constructs into endothelial cells in vivo. Nanoparticle-mediated gene delivery of STAT3 restored endothelial proliferation and stimulated lung angiogenesis in neonatal S52F Foxf1 mice, supporting efforts to treat ACDMPV by stimulating STAT3 signaling.

We will use S52F-Foxf1 mice to determine the delivery routes and dosage of FOXF1-stabilizing small molecule compound in order to improve the vascular development and survival rates in ACDMPV mice."

NORD Request for Proposals ("RFP") – 2018 Research Grant:

NORD intends to issue 2018 RFPs in May. The ACDA has not yet officially released an announcement regarding the 2018 NORD grant opportunities but please be on the lookout for updates from NORD in May if you are interested in potential scientific research studies and/or clinical research studies related to ACDMPV.

Role of Serotonin Transporter in Human Lung Development and in Neonatal Lung Disorders:

The ACDA recently reviewed the research paper entitled, “The Role of Serotonin Transporter in Human Lung Development and in Neonatal Lung Disorders” by E.C.C. Castro, P. Sen, W.T. Parks, C. Langston and C. Galambos as published in Canadian Respiratory Journal in 2017 (Full Article and citation). The study contemplates a new area of exploration in both the pathogenesis of and possible future development of new therapeutic strategies for ACDMPV. In the meantime, the next proposed step of this research is to confirm whether or not ACDMPV patients display elevated serotonin levels.
AWARENESS NEWS

Update on Rare Disease Day (February 28, 2018):

We are pleased to announce $4,219.32 was raised by the ACDA and an additional £1,026 was raised by our sister UK registered charity, The David Ashwell Foundation, for a grand total of $5,631 in connection with Rare Disease Day on February 28, 2018!! 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!

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

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

- Lee County Bank  
  (Fort Madison, IA)  
- Family Design Resources  
  (Harrisburg, PA)  
- Discover Little Miracles 
  (St. Michael, MN)  
- East Quogue Teachers Association  
  (East Quogue, NY)  
- East Quogue Elementary School 
  (East Quogue, NY)  
- The Suffolk County District Attorney, Economic Crime & Cyber Crimes Bureau  
  (Hauppauge, NY)  
- Suffolk County Department of Law & Suffolk County Office of Risk Management  
  (Hauppauge, NY)  
- Suffolk County Office for the Aging  
  (Hauppauge, NY)  
- Suffolk County Budget Office & Suffolk County Federal & State Aid Unit  
  (Hauppauge, NY)  
- Suffolk County Department of Audit & Control & Suffolk County Office of Administration  
  (Hauppauge, NY)

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

- Teesside University  
- Human Nutrition Research Centre, Newcastle University  
- Appleton Wiske School  
- Kirby Great Broughton school  
- Lewes priory school  
- Moulsoomb primary school  
- Hertford infants school

Please click HERE to check out our ACDA families and friends wearing the ACDA logo and #JeansforGenes on Rare Disease Day!
The ACDA created flyers for use at workplaces and schools encouraging colleagues and classmates to donate $5 / £5 / €5 to wear jeans to work or school on February 28 for Rare Disease Day. The flyers can be modified for a Jeans Day at any time of year, contact the ACDA for help!

Below please find a “Thank You” badge for donations made on Rare Disease Day 2018 in your child’s honor:
**BBC Tees radio show (David Ashwell):**

Amelia Ashwell, founder of The David Ashwell Foundation and ACDA mother to David (March 4, 2011 – March 19, 2011), was on the BBC Tees radio show with Mike Parr on February 28, 2018! Amelia spoke about Rare Disease Day and how having a rare disease diagnosis for David changed her family’s life. She also spoke about UK lung transplant recipient, Imogen Bolton. [Listen](#) to Amelia at 2:31:31 into the program.

---

**ACDA tattoo (Shelby Myers):**

The youngest daughter of Madonna Myers, ACDA founder and mother to Shelby (May 31, 1996 – July 3, 1996), recently got a tattoo of the ACDA logo to remember the sister she never knew. What a beautiful tribute between siblings.

---

**ACDA connections (Sadie McCasland and Joey Eschweiler):**

Candice McCasland, ACDA mother to Sadie (February 4, 2014 – February 19, 2014), recently met Emily Eschweiler, ACDA mother to Joey (January 10, 2010 – January 23, 2010), during Emily’s family trip to Florida. Emily and Candice are both members of the ACDA Communications Committee and this is the sixth ACDA family Candice has met in person!

---

**International Bereaved Mother’s Day (May 6, 2018):**

[International Bereaved Mother’s Day May 6, 2018](#)
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

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

Update from NORD:

The NORD balance of the Research Fund (ACD) as of April 23, 2018 is $xxxx.

Balance of ACDA account:

The balance of the ACDA bank accounts as of April 30, 2018 is $xxxx.

Update from AmazonSmile:

The ACDA was issued a $xxxx donation from the AmazonSmile Foundation as a result of AmazonSmile program activity between October 1 and December 31, 2017. 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

Update from Spreadshirt:

The accrued commission payment from Spreadshirt between January 23, 2018 and April 25, 2018 is $xxxx. Our Spreadshirt store featuring the ACDA logo has sold 153 items since opening in September 2015! Items with the ACDA logo are available for purchase in our Spreadshirt store HERE. The ACDA earns a commission equal to 20% of every product sold. Look for a free shipping event on May 4 – 6, 2018! (Use code: SHIP2ME)

Donations Received:

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

<table>
<thead>
<tr>
<th>Family/Friend Name</th>
<th>Amount</th>
</tr>
</thead>
<tbody>
<tr>
<td>Smith</td>
<td>$250</td>
</tr>
<tr>
<td>Johnson</td>
<td>$150</td>
</tr>
<tr>
<td>Rodriguez</td>
<td>$100</td>
</tr>
<tr>
<td>Garcia</td>
<td>$200</td>
</tr>
<tr>
<td>et al.</td>
<td>$350</td>
</tr>
<tr>
<td>ALVEOLAR CAPILLARY DYSPLASIA</td>
<td>ACDA Notes</td>
</tr>
<tr>
<td>-----------------------------</td>
<td>------------</td>
</tr>
</tbody>
</table>

### Table of Contents

- Introduction
- Pathophysiology
- Diagnosis
- Treatment
- Prognosis
- Outcome
- Conclusion

### Key Points

- Alveolar capillary dysplasia (ACDA) is a rare genetic disorder characterized by abnormal development of capillaries and alveoli in the lungs.
- The condition is typically inherited in an autosomal recessive pattern.
- Diagnosis is made through genetic testing and imaging studies.
- Treatment options include supportive care and lung transplantation in severe cases.
- Prognosis varies depending on the severity of the condition, with many cases leading to early infant mortality.
- Research is ongoing to develop new treatments and improve outcomes.

### Further Resources

- [Alveolar Capillary Dysplasia Association](https://www.acda.org)
- [Genetic Counselors](https://www.ncbi.nlm.nih.gov/pubmed)
- [Clinical Trials](https://clinicaltrials.gov)
Pottery fundraiser (Johnny Rista):  

John and Eliza Rista, ACDA parents to Johnny (February 20, 2013 – March 4, 2013), organized a fundraiser for ACDMPV research in February 2018. A local business, Meg-Art Pottery Painting Studio, generously agreed to donate 20% of all their pottery sales on February 11, 2018 to the ACDA.

The Ristas organized this event as a fun way to raise awareness and funds for the ACDA in honor of Johnny’s fifth birthday. There was a tremendous turnout and over $1,000 was raised for ACDMPV research.

Banners:

If you would like to request use of a banner in the United States, please send an email to president@acdassociation.org to discuss sign-up for availability and shipping information.

Volunteer Grants:

To honor those who give their time and service to causes important to them, Bank of America, NA, through its Bank of America Charitable Foundation, awards unrestricted grants for volunteer work to any eligible nonprofit organization for which an employee has committed substantial volunteer hours within a calendar year. For 50 hours of volunteer time within a calendar year, Bank of America Charitable Foundation will give a $250 grant. For over 100 hours of volunteer time within a calendar year, the grant is $500.

Thank you to Bank of America, NA for providing a $500 grant to the ACDA in honor of John Rista’s volunteer work in 2017 for the ACDA. Please let us know if similar volunteer grants are offered through your employer!

*We are sorry we do not know the child for whom the honorary contribution was made. Please contact us to let us know.
REPORT FROM STICHTING ACD (NETHERLANDS):

So many amazing events are occurring in the Netherlands, as led by Mathijs Lourens, ACDA father to Myla (November 20, 2015 - December 17, 2015), including marathons, video reports, banners, newspaper articles, television interviews, speaking engagements and more. Please read all the details below!

**Radboudumc (March 6, 2018):**

On March 6, 2018, Mathijs gave a one hour lecture to a group of young medical students at Radboud University Medical Center in Nijmegen to teach the next generation of medical professionals about ACDMPV.

**Rotterdam Marathon (April 8, 2018):**

Mathijs ran the Rotterdam marathon on April 8, 2018 for the second year in a row in support of his Dutch ACDMPV foundation, “Stichting ACD.”

In Mathijs’ own words before the marathon, “Thank you all for listening and giving me the chance to share our adventure over here in the Netherlands. Hopefully we meet someday! I would like to give a special shout out to Roelina Jut, ACDA mother to Jasmijn (February 8, 2013 – February 23, 2013), for helping me so much with translating and designing the logo for the Team Myla running shirts! Myla’s picture will be waiting for me around 41K, held by friends and Jessica and the kids will be there as well to support all runners! There will be 15 runners wearing a Team Myla t-shirt! That makes me proud! There was a special fundraiser for Stichting ACD in Bahrein, organized by an old classmate who lives there. He will run the marathon as well! …We’re raising awareness for ACD and like I promised before: We’ll continue doing that for as long as needed.”

After completing the Rotterdam marathon, Mathijs shared, “Magic happens all the time.
Once again it was a fantastic day! More info soon, but I wanted to share this special picture. **40,000 people saw the picture below** on the marathon route at 5K when everyone was still fresh.”

![Picture](image1)

Please be sure to check out the [video report](#) Team Myla made during the April 2018 Rotterdam marathon! In addition, Team Myla also recently shared a separate [video](#) report from the October 2017 Amsterdam marathon (see Issue #58 of ACDA Notes).

**National Newspaper Article (April 3, 2018):**

Please click [HERE](#) to read (in Dutch) about Myla’s story and Mathijs’ remarkable contributions, as published on April 3, 2018 in *Algemeen Dagblad (AD)*, a national Dutch daily newspaper based in Rotterdam, Netherlands.

![Image](image2)

**National Television Report (April 6, 2018):**

Please click [HERE](#) to watch (in Dutch) a television interview featuring Mathijs, as recorded on April 6, 2018 with a national news network in the Netherlands. In addition, the interview highlights Professor Tibboel of Erasmus MC-Sophia Children's Hospital in Rotterdam for his expertise on ACDMPV. Take a look and you’ll see Professor Tibboel reviewing the updated ACDA brochure (now available in [Dutch](#)).

**Article published by Erasmus MC-Sophia Children's Hospital (April 23, 2018):**

Please click [HERE](#) to read (in Dutch) an article published by Erasmus MC-Sophia Children's Hospital in Rotterdam.

![Image](image3)

Mathijs Lourens, ACDA father to Myla (November 20, 2015 - December 17, 2015) presenting the ACDA brochure (translated in Dutch!) to Professor Tibboel of Erasmus MC-Sophia Children's Hospital in Rotterdam.

**Looking ahead:**

Mathijs is already working on future plans to expand his marathon adventure across the border. He is considering running an upcoming Cologne marathon as there is an ECMO center in Mannheim, Germany, in the hopes of bringing ACDMPV awareness to Mannheim as well. **Please contact Mathijs if you are interested in participating in his upcoming plans to raise awareness for ACDMPV in the Netherlands or Germany.**
REPORT FROM THE DAVID ASHWELL FOUNDATION (UK):

The past few months has reminded me that there is a lot of good will and kindness in the world. Sometimes you just have to ask and there can be amazing results. But also that we need to be better organised and get the rare disease day requests to schools in June 2018 for Feb 2019!! So if you know a school, let’s get it into their planning diary well in advance.

The David Ashwell Foundation is run by Simon and myself, we are so very grateful to anyone who uses the charity or donates to the charity in memory of their precious babies. Thank you especially to those who have a standing order and put money into The David Ashwell Foundation charity bank account each month (ACD and non ACD families).

Since March 2011 £197,488 ($271,978) has been raised for ACD Research by families in the UK and Europe. This has contributed to the funding raised internationally to the ACDA. Since April 2017 we have raised £16,161 ($22,169) which is an incredible amount of money given how few we are! This will be added to the ACD research fund at NORD ready for the next research grant.

It’s been a busy time for us with Rare Disease Day, the ceilidh and the awful weather we had in the UK! I did a BBC Tees (local radio) interview about rare disease day and David (as the snow started!). On Rare Disease day I was due to give a public lecture about having a child diagnosed with a rare disease. Writing the presentation was emotional, but after all the preparation it was cancelled as the University shut due to the excessive amount of snow we had. On the same (snow) day we had a cake sale and despite the fact that there were very few staff or students on campus we still raised £106.00. Two schools local to us raised money on rare disease day – again both were postponed as the schools were shut due to the bad weather! Appleton Wiske raised £74 and Kirby Great Broughton (Auden’s school) raised £197. Online, our collection for rare disease day raised £449.50 this included donations via Imogen’s incredible big sister Aliessia (who is a complete superhero!). Via Hayley (Imogen’s mum) there were rare disease day contributions from three schools Lewes Priory, Moulsoomb Primary and Hertford Infants. Our co-operative supermarket collected change at the end of the tills for the month of March which raised £200. A rare disease day total of £1,026 – (given the weather – this is amazing!)

Amelia and Simon Ashwell with compère, Mike Charlton, at the ceilidh on March 3, 2018, which raised an amazing £3,564 for ACDMPV research!

This year, with the bad weather and the impact it had on life in general I have to say that organizing the ceilidh nearly put me over the edge!! My mum couldn’t make it here to help with the final bits and pieces but my incredible sister Anisa made it from Dubai (despite the snow) plus so many friends mucked in. The weather had a big impact – it was a lot quieter than previous years. It was
still a fun night and an incredible total of £3,564 was raised.

Amelia and Simon Ashwell in action at the ceilidh!

This included £457.00 raised by our small co-operative food store on the Saturday of the ceilidh. The staff took turns on the static bike in store with a big bucket and raised an incredible amount. The following week they ran a bonus ball game and raised an additional £60. In addition Pat and Jane who both work in the store organized a sponsored walk and raised another £391!! These ladies – who I know because I meet them in the shop are an incredible force and have generated so much money for our charity. On Anisa’s flight to Newcastle the crew collected money (in all kinds of currency) and donated it for the charity. There is a lot of kindness in the world!

https://www.thegivingmachine.co.uk/causes/the-david-ashwell-foundation/support/

All UK & Europe based families are welcome to use The David Ashwell Foundation as a means of fundraising for ACDMPV Research. Please raise awareness of our small charity via social media including twitter and Facebook.

We are more than happy to hear from other families who would like to use the charity to raise money for ACDMPV Research. Simon and Amelia run The David Ashwell Foundation on a voluntary basis. All money raised will be transferred to NORD to fund ACDMPV Research.

Thank you to all who have used The David Ashwell Foundation to fundraise in memory of their precious child.

For additional information, please contact Simon and Amelia.

Website: http://David Ashwell Foundation
Email: davidashwellfoundation@yahoo.co.uk
Twitter: @TDavidAshwellF
Mobile (Amelia): 07855473686
Welcome to New Families

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

ACDA COMMITTEE POSITIONS

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
Safe Arrivals!

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

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)