ACDA
ALVEOLAR CAPILLARY DYSPLASIA ASSOCIATION
acdassociation.org
WINTER 2018
Issue 62

ACDA NOTES

FROM THE ALVEOLAR CAPILLARY DYSPLASIA ASSOCIATION

What's INSIDE

2017: Year in Review

As we take a look back at 2017:

- **Twelve new families registered with the ACDA. These families are from the USA, Australia, UK and Canada.**
- **Two additional presumptive prenatal diagnoses of ACDMPV without a family history.**
- **One NORD research grant in the amount of $50,000.**
- **One NIH R01 research grant in the amount of $1,900,000.**
- **Nineteen rainbow babies born to ACDA registered families.**
- **One additional tattoo of the ACDA logo.**
- **More than fifteen journal articles published about ACDMPV.**
- **Numerous successful fundraisers for ACDMPV research and awareness events.**
- **One continued goal – To find the cause of and cure for ACDMPV.**

Regards, Eliza Rista, President

FEBRUARY 28, 2018 IS RARE DISEASE DAY

Rare Disease Day is an international advocacy day to bring widespread recognition of rare diseases as a global health challenge. The day is celebrated on the last day of February every year – February 28 in 2018. The ACDA has signed on once again to partner with NORD to support this awareness campaign and we encourage everyone to participate in some way. **Click to see how the ACDA supported Rare Disease Day in 2017 and 2016!** Click HERE for ways to participate in the U.S. in 2018 and click HERE to visit the EURORDIS website for Rare Disease Day 2018 in Europe.

Below are a few suggested ideas:

- Click HERE ($), HERE (£) or HERE (€) to print a flyer for anyone to use at their workplace that encourages employees to donate $5 / £5 / €5 to the ACDA and wear jeans to work on Wednesday, February 28 for Rare Disease Day.
- Join a social media "rare disease day" campaign by changing your profile and cover pictures to share the importance of rare disease awareness! Click HERE to upload a photo and show how much you care about rare! Use the hashtags **#RareDiseaseDay**, **#WRDD2018**, **#ShowYourRare, #ACD**, **#ACDMPV**
- Organize a fundraiser to raise money for the next ACDMPV research grant.
- Contact your local newspaper to write an article about what Rare Disease Day means to you. Click HERE to read various articles previously written about our ACDA families.

RESEARCH NEWS

2017 NORD Grant Update:

It is our great pleasure to share the previously announced 2017 NORD grant in the amount of **$50,000 for ACDMPV research** (see Issue #59 of ACDA Notes) **was recently awarded to 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)*."

The ACDA is thrilled the Cincinnati team has been awarded this ACDMPV research grant. The genetic research team at Baylor has worked in direct collaboration with the developmental biology research group at Cincinnati, including publishing together an article about FOXF1 overexpression (see Issue #57 of ACDA Notes). Dr. Pradhan (the 2017 grant recipient) is a member of the Kalinichenko Research Lab at Cincinnati. The long-term goal of the Kalinichenko Research Lab is "to discover novel therapeutic approaches and generate novel FDA-approved drugs for treatment of these severe respiratory disorders." To learn more about the Kalinichenko Research Lab, please click HERE and read about their current projects.

Funding for NORD grants in support of ACDMPV research is raised through the hard work, contributions and fundraising efforts of families and friends affected by ACDMPV. THANK YOU for making these very important research grants possible.

Atypical ACDMPV and lung transplants in St. Louis from 1998-2016:

Several significant ACDMPV research institutions recently collaborated together to publish a journal article entitled, *"Infants with Atypical Presentations of Alveolar Capillary Dysplasia with Misalignment of the Pulmonary Veins Who Underwent Bilateral Lung Transplantation*," as published in The Journal of Pediatrics in November 2017 and found HERE. The collaborators include **Cincinnati** Children's Hospital Medical Center in Cincinnati, Ohio, USA, **St. Louis** Children's Hospital in St. Louis, Missouri, USA, **Baylor** College of Medicine in Houston, Texas, USA and **Northwestern** University Feinberg School of Medicine in Chicago, Illinois, USA.

Our thanks to Dr. Simon Ashwell, ACDA father to David (March 4, 2011 – March 19, 2011), for the following summary:

This paper reports the details of 6 infants with atypical ACDMPV who had lung transplantation at St Louis Children's Hospital over an 18-year period (1998-2016) and compares them to infants with classical ACDMPV. Infants with classical ACDMPV have progressive, severe breathing problems within hours of birth. By contrast the infants with atypical ACDMPV developed breathing problems 2-7 months after birth. Of the 6 transplanted infants, 3 were alive at the time of the report, 2 with normal lung function and 1 with chronic rejection (bronchiolitis obliterans). Of the 3 infants who died one required a second lung transplant at 5 years of age due to bronchiolitis obliterans but died due to progressive kidney failure 2 months later; one died of bronchiolitis obliterans at 9 years of age; the other infant died around 3 months after transplant at 18 months of age due to recurrent unexplained breathing problems. A large, international registry of childhood lung transplantation for all causes reported an average survival after lung transplant of 5.3 years, with infants having a slightly better average survival of 6.4 years.

Infants transplanted due to ACDMPV therefore have a similar outcome to those transplanted for other reasons.

Blood was available for genetic testing in 5 of the 6 transplanted infants. 2 had de novo FOXF1 mutations that had not previously been reported; one had a deletion close to FOXF1; No mutations or deletions were found in the remaining 2 infants. As one had a sibling with ACDMPV it is clear that other genes that those current recognised must contribute to ACDMPV.

The removed lungs from the transplanted infants showed characteristic microscopic changes of ACDMPV but these were focal or patchy compared with the diffuse and extensive changes in the lungs of infants with classical ACDMPV. This finding makes diagnosis of ACDMPV by (single specimen) lung biopsy potentially challenging as there are areas of lung with normal tissue. As 80-90% of infants with ACDMPV have FOXF1 mutations or local deletions, this suggests that the use of genetic testing may prevent the need for lung biopsy. There was no correlation of the microscopic changes in the lungs with the severity of the breathing problems or genetic abnormalities.

This paper shows that infants with atypical ACDMPV have patchy microscopic changes in the lungs, leading to delayed presentation with breathing problems compared to infants with classical ACDMPV. Such infants have a similar outcome following lung transplant to infants transplanted for other conditions.

AWARENESS NEWS

The ACDA is excited there are a number of ACDMPV awareness events to summarize, including a research presentation, NORD spotlight, newspaper article, new ACDA informational brochure, upcoming Rare Disease Day 2018 events and several personal tributes, each as described below.

Presentation (ASHG 2017)

The 67th annual meeting of the American Society of Human Genetics (ASHG) was held in Orlando, Florida on October 17 – 21, 2017. ASHG 2017 featured invited presentations by the world's leading geneticists, in addition to symposia, workshops and abstract-driven sessions about new developments in basic, translational and clinical human genetics research and technology. Dr. Pawel Stankiewicz from Baylor College of Medicine was in attendance to talk about FOXF1, specifically "*Evolutionarily young LINE elements initiate recurrent DNA breaks forming different-sized CNVs via both NAHR and microhomology-mediated DNA replication mechanisms*." Click HERE to read the abstract.

Rare Disease of the Day (NORD):

On October 16, 2017, ACDMPV was featured as the Rare Disease of the Day at the National Organization for Rare Disorders (NORD). October 16, 2017 was also the first day of the #NORDSummit2017 in Washington, DC with 670 passionate individuals committed to improving the lives of people with #rarediseases. Thanks for the support NORD!

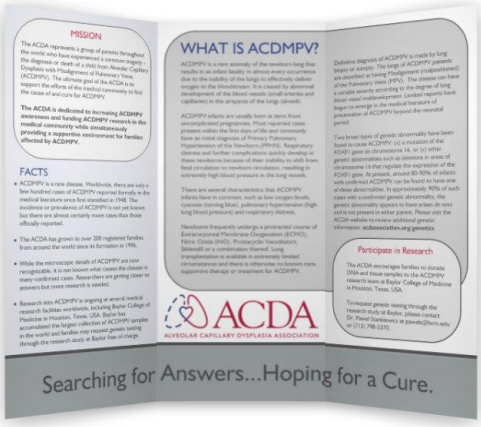
Families in the News (Valerie Lihs):

Below is an article in honor of **Valerie Rose Lihs** (April 19, 2017 – May 30, 2017):

Two families will benefit from DEC Hands event
Fort Madison Daily Democrat
November 3, 2017
Fort Madison, Iowa, USA

ACDA Brochures (New!!):

The updated ACDA brochure **has a wonderful new look**! View, save, email or print your own copies of the ACDA brochure; click for **ENGLISH**, **DUTCH** or **ITALIAN**.

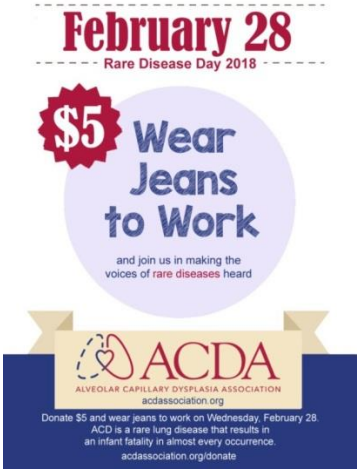


The ACDA extends its appreciation to **Mathijs Lourens** and **Roelina Jut** for the Dutch translation and **Federica di Paolo** for the Italian translation. *Please let us know if you are available to translate to other languages for addition to our website*.

Wear Jeans to Work Flyers:

As referenced on page 2, **the ACDA has created flyers for anyone to use at their workplace that encourages employees to donate $5 / £5 / €5 to the ACDA and wear jeans to work on Wednesday, February 28 for Rare Disease Day.** Click HERE (**$**), HERE (**£**) or HERE (**€**) to print or email the full size images. We hope you will consider asking your workplace to participate! This was a terrific awareness event and fundraiser last year on Rare Disease Day.

Donations can be submitted individually or collectively at **acdassociation.org/donate** and we are happy to track the amounts donated in your child's honor.



ACDA headstone (Dawson Nye):

Brittany Nyes, ACDA mom to Dawson (June 8, 2017 – July 27, 2017), included the ACDA logo on the back of Dawson's headstone. It is absolutely beautiful and the ACDA is deeply honored to be a part of Dawson's very special tribute.

ACDA tattoo (Valerie Lihs):

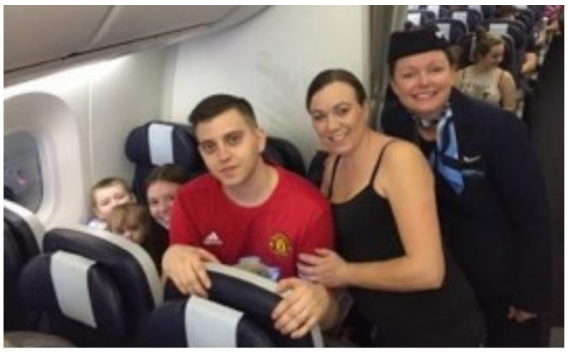
Lisa Lihs, ACDA mom to Valerie (April 19, 2017 – May 30, 2017), recently got an amazing tattoo in honor of Valerie that included the ACDA logo.

Surprise for Imogen Bolton:

The following is written by Jo Taylor, ACDA mum to Alex (May 8, 2011 - May 17, 2011) and James:

In October 2017, **I had the pleasure of meeting Imogen**, one of our ACDMPV warriors. The ACDA previously introduced you to Imogen as an ACDMPV survivor in the UK and Europe's smallest lung transplant recipient (see Issues #56, #58 and #61 of ACDA Notes).

Imogen's mum, Hayley, had told me months before that she had booked a holiday with my airline company, so I asked my managers if I could be put on their flight.



I also tried to swap with the crew on it as I wasn't rostered it and used that opportunity to tell the crew Imogen's story.

As many of you know, **my work raised £36000 from on-board collections in 2012/2013 so the majority of crew knew how much of a miracle it was to have a little one onboard who'd had a double lung transplant**.

One of the crew suggested that we put a box in the crew room for donations for Imogen and her friends travelling.

I also contacted the Sales department to see if we could get **six pilot bears** for all the children. I used to work with the Sales Manager years ago so filled him in on Alex's story too and he kindly sent them.

On the morning of the flight, I told Hayley that I was going to be on her flight and to call if any hassles with Security and getting Imogen's medication through. Luckily all was ok and I arranged for the boarding gate to let Imogen's party board first so we could take photos and let them get settled.

In the crew room, I'd bought in bags to make up '**goody bags**' for everyone and my colleagues were amazing again, donating sweets and stickers so I could distribute.

Gordon, who was supposed to be the Cabin Manager had also off his own back, made up 6 bags for everyone too. Luckily for us all, Gordon was working down the back of the plane where Imogen's family were sat and so both of us could look after Hayley and everyone!

I'd found out where Hayley was sitting and so picked my position accordingly so I was near them and could serve them. It was only a short flight to Majorca, but it was an honour to meet everyone and have a good catch up with Hayley.

Once we'd landed, I also had arranged with the Captain to let them all visit the 787 Dreamliner flight deck and have photos taken.

I'm sure that they all had a wonderful holiday and **it truly was an honour to get to meet a real life miracle gorgeous girl** along with her lovely well-mannered brother & sister as well as Hayley and Jason and help make some memories!

UPDATE FROM "STICHTING ACD" IN THE NETHERLANDS:

Stichting ACD celebrated its one-year anniversary on January 18, 2018!

Mathijs Lourens, ACDA father to Myla (November 20, 2015 - December 17, 2015), ran the Amsterdam marathon on October 15, 2017 in support of his Dutch ACDMPV foundation, "**Stichting ACD**."

Next, Mathijs is running the Rotterdam marathon on April 8, 2018! **Please contact Mathijs if you would like to cheer for Team Myla during the Rotterdam marathon to raise awareness for ACDMPV**. The ACDA sends our very best to Mathijs and the team of Stichting ACD runners in Rotterdam. We look forward to providing pictures and an update in the next newsletter!

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 December 31, 2017 is $[redacted]. The minimum amount required for the issuance of a 2018 NORD grant is $35,000 (increased from $33,500 effective as of the 2017 grant cycle).

Balance of ACDA account:

The balance of the ACDA bank account as of January 25, 2018 is $[redacted].

Update from Spreadshirt:

The accrued commission payment from Spreadshirt between October 13, 2017 and January 22, 2018 is $[redacted]. Items with the ACDA logo are available for purchase in our Spreadshirt store HERE.

Update from AmazonSmile:

The ACDA was issued a $[redacted] donation from the AmazonSmile Foundation as a result of AmazonSmile program activity between July 1 and September 30, 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

Donations Received:

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

[redacted]	[redacted]
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[redacted]

**We are sorry we do not know the child for whom the honorary contribution was made. Please contact us to let us know.*

#GivingTuesday Update:

WOW. The ACDA's fundraising goal for #GivingTuesday on November 28, 2017 was $500 and, with your help, we were able to bring in just over **$1,600**!!

One day, one mission, one BIG thank you. We give our thanks to those who donated to the ACDA on #GivingTuesday. The money raised will be contributed directly to ACDMPV research and supports the efforts of the medical community to find the cause of and cure for ACDMPV.

Thanks very much for the generous #GivingTuesday donations in honor of *Sadie McCasland, Xzendria Swenson, Amelia Weaver, Avery Spicer, Fraser Dunne, Joey Eschweiler, Johnny Rista, Ronan Bush, Phoebe Bush, Fallon Rilling, Dawson Nye* and *Ainsley Abodeely*.

Our gratitude extends not only to our amazing donors but to each and every person who shared a post or email to spread awareness, asked their family and friends to consider a donation or helped in any possible way to make our #GivingTuesday campaign a huge success in 2017. THANK YOU!!

Giving Bean:

The ACDA held our third annual **Giving Bean coffee fundraiser** during the 2017 holiday season!

In-person sales (40%): Please note we can re-open our in-person sales campaign at any time there is an interest in selling coffee for a fundraiser; just let the ACDA know! In-person sales are fun to do (just like selling Girl Scout cookies) and 40% of sales go to ACDMPV research. Additionally, if you sell more than 50 bags through in-person sales, your bags will come with the ACDA logo affixed on the bag.

Online sales (25%): As an alternative to the in-person sales option, please click HERE at any time to go directly to the ACDA supporter page and 25% of any purchases made through such link will go to the ACDA.

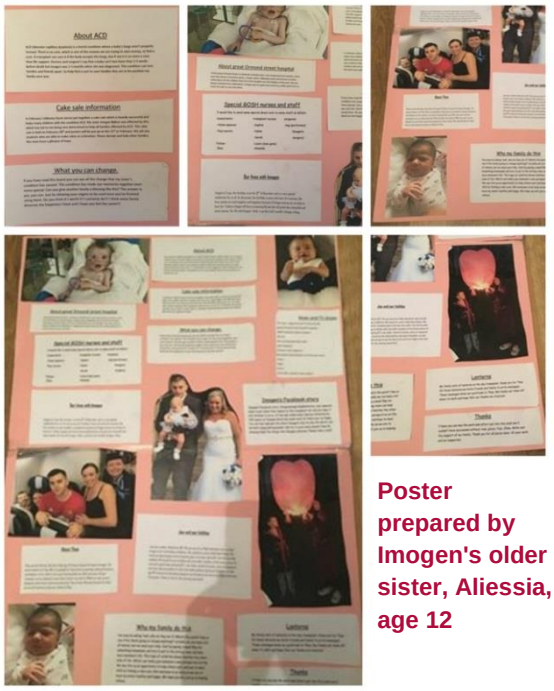
Giving Bean's flavor selection is vast and delicious and averages $12 a bag, which makes it a great gift at any time of the year and a wonderful way to raise funds for ACDMPV research.

Bravelets™:

Don't forget about our Bravelets™ fundraiser! Bravelets™ sells bracelets for profit in the U.S. with a mission to encourage people to be brave during hard times. 10% of each item purchased through this LINK will be donated to the ACDA.

School Fundraiser (Imogen Bolton):

Aliessia, age 12, daughter of ACDA mom Hayley Bolton and older sister of lung transplant recipient Imogen Bolton, independently organized a fundraiser at her school to raise funds for ACDMPV research. She made and sold little hot chocolate cones and reindeer food and her friends made cakes to sell. Aliessia also knitted scarfs to sell and created a massive poster explaining all about ACDMVP and Imogen's story. **She raised £115 ($157)!!** The ACDA is so incredibly proud of this young lady's hard work and dedication to ACDMPV research!



Poster prepared by Imogen's older sister, Aliessia, age 12

REPORT FROM THE DAVID ASHWELL FOUNDATION:

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 (ACDMPV and non ACDMPV families).

Since March 2011 **£190,447 ($261,640)** has been raised for ACDMPV Research by families in the UK and Europe. This has contributed to the funding raised internationally to the ACDA.

We were so thrilled to hear about Imogen's big sister Aliessia's cake sale. Independently, Aliessia organised a sale in her school and produced information boards about ACDMPV. She raised £115 ($157). This was such amazing initiative by Aliessia who is only 12 years old. She is an inspiration to us all! In November I held a 'virtual' Neals Yard event and raised £150 ($206).

I am busy getting things in motion for our annual Ceilidh for David on 3rd March 2018 – if any local (ish) families would like to attend or help please let me know.

As ever, can I plea with all UK families to use the **Giving Machine** to generate income from online shopping. Everyone in the UK can do this – it is so easy to register and is a way of earning easy and free money. Please also encourage your friends and family too!

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

UK families you can set up direct debits to The David Ashwell Bank Account (we have a number of people who do this).

Additionally we have a Virgin Money Giving account where you can set up a page in memory of your child or for special occasions (such as birthdays or Christmas).

If you live in the UK (and elsewhere), there are a number of options available for funding ACDMPV research through The David Ashwell Foundation.

1. You can make a donation directly, using the Virgin Money giving website to gather donations for your fundraiser. http://David Ashwell Foundation
2. Fundraise while you online shop (The Giving Machine) (a percentage of what you spend is donated)
3. Fundraise when you ebay (ebay for Charity).
4. Collect postage stamps http://David Ashwell Foundation

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

Remembering Our Babies

[redacted]

[redacted]

Welcome to New Families

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

- [redacted]

(*continued on following page*)

[redacted]

Safe Arrivals!

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

[redacted]

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)