

ACDA NOTES

FROM THE ALVEOLAR CAPILLARY DYSPLASIA ASSOCIATION

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This newsletter is packed full of information because a lot has happened since the last ACDA Notes! A few highlights:

1. **The 2015 NORD research grant for \$50,000 was issued to Dr. Partha Sen at Northwestern University (see page 2 for details) and another \$50,000 NORD grant is expected to be issued in 2016 (see page 5 for details);**
2. **Dr. Pawel Stankiewicz at Baylor College of Medicine had a new research article recently accepted for publication in the medical journal, Human Genetics (see page 2 for details);**
3. **More than \$1,200 was raised by families and friends of the ACDA in connection with Rare Disease Day on February 29, 2016 (see page 3 for details);**
4. **Three newspaper articles were published about ACDA registered families (see page 4 for details);**
5. **The ACDA has two new custom banners for use by ACDA registered families at fundraising events (see page 5 for details);**
6. **The David Ashwell Foundation report (see page 9 for details); and**
7. **Four additional ACDA committee members were approved in January 2016 (see page 11 for details).**

On behalf of the ACDA, thanks to everyone for all of the dedicated work in support of our very important mission!

Regards,
Eliza Rista, Secretary

2015 NORD Research Grant Recipient

The ACDA is pleased to announce NORD's Medical Advisory Committee recommended, and the NORD Board of Directors approved, the awarding of a grant for ACDMPV research in the amount of **\$50,000** (with funding raised in coordination with the ACDA and The David Ashwell Foundation) for the 2015 grant cycle. The grant was distributed as follows:

Partha Sen, PhD (primary investigator) and Aaron Hamvas MD (co-investigator)
Northwestern University

Title: *Characterizing the FOXF1 gene network in lung development*

The aim of this two year grant is to help understand the role of FOXF1 in ACDMPV and in lung development, along with the development of other organ systems. The overall objective is to understand the genetic basis of ACDMPV that may lead to therapeutic developments of this deadly disorder.

Please visit the ACDA website (<http://acdassociation.org/research-grants>) for complete information about current and prior NORD research grants, including the status of the current 2016 grant cycle (see also Page 5 of this newsletter).

RESEARCH NEWS

Human Genetics:

In March 2016, Dr. Pawel Stankiewicz at Baylor College of Medicine informed the ACDA that the paper "*Pathogenetics of Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins*" was recently accepted for publication in the medical journal, Human Genetics. It was a huge amount of work and updates current knowledge about the genetics of ACDMPV based upon past research and new findings from recent work. Baylor has accumulated the largest collection of ACDMPV samples worldwide (N=141 families), in which they have identified 86 pathogenic variants in the FOXF1 locus: 38 deletion CNVs, a complex rearrangement and 47 point mutations. DNA was not of sufficient quality for genetic testing in most of the remaining 55 families. The paper demonstrates the complexity of genomic and epigenetic regulation of the FOXF1 gene in 16q24.1.

The ACDA has asked Dr. Simon Ashwell, father to David (March 4, 2011 - March 19, 2011), for his assistance in preparing a summary of this important article upon its publication in Human Genetics. Simon's summary will initially be posted on the ACDA website (<http://acdassociation.org/journal-articles>) as soon as available and will also be included in a subsequent ACDA quarterly newsletter.

PhD Thesis:

In October 2015, Avinash Vijay Dharmadhikari, graduate student and mentee of Dr. Pawel Stankiewicz, submitted his dissertation (thesis) entitled "*Essential Role of Mesenchymal FOXF1 Dosage in Lung Maturation*" to the faculty of the Graduate School of Biomedical Sciences at Baylor College of Medicine. Avinash has investigated the role of FOXF1 since 2011. Please contact the ACDA if you would like to request a copy of his thesis.



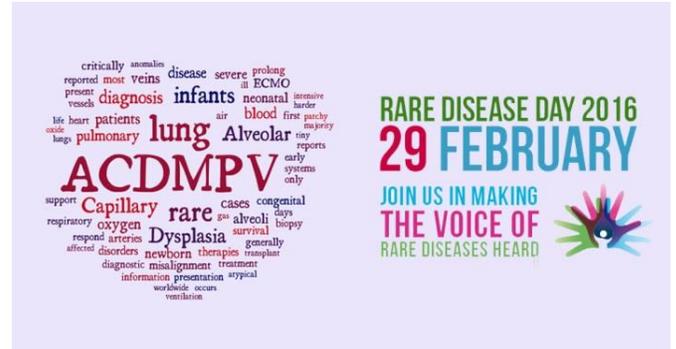
AWARENESS NEWS

The ACDA is excited to report there is a lot of ACDMPV awareness news to summarize, including research presentations, Rare Disease Day events, newspaper articles and proclamations as described below.

Presentations:

- The 13th International Congress of Human Genetics (ICHG2016) was held in Kyoto, Japan on April 3 – April 7, 2016. The ICHG meeting has been held every 5 years since 1956. Dr. Pawel Stankiewicz at Baylor College of Medicine was in attendance to talk about FOXF1, specifically “*Unraveling the role of genomic imprinting at 16q24.1 in pathogenetics of alveolar capillary dysplasia with misalignment of pulmonary veins and maternal uniparental disomy 16.*” Click [HERE](#) to read the abstract.
- The European Society of Human Genetics (ESHG2015) held its European Human Genetics Conference in Glasgow, Scotland, United Kingdom on June 6-9, 2015. Avinash Dharmadhikari at Baylor College of Medicine was in attendance to talk about FOXF1, specifically “*Does paternal imprinting of FOXF1 on 16q24.1 explain the maternal UPD(16) phenotype?*” Click [HERE](#) to read the Abstract (C07.1) and [HERE](#) to read the presentation materials.

Rare Disease Day:



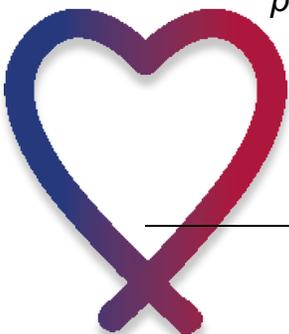
More than \$1,200 was raised by families and friends of the ACDA in connection with Rare Disease Day on February 29, 2016!! We would like to give our special thanks to the employees of Suffolk County District Attorney’s Office (New York), Smithtown Library (New York), Family Design Resources (Pennsylvania), Mercer County Children and Youth Services (Pennsylvania) and Pleasant Lea Elementary (Missouri) for their outstanding support of our ACDA families as part of a “\$5 to Wear Jeans to Work” fundraiser on Rare Disease Day. Our thanks to the families of Fallon Rilling, Callie Murray and Finley Smith for their help in coordinating this event with their employers.

February 29

Rare Disease Day 2016

\$5 Wear Jeans to Work

and join us in making the voices of rare diseases heard



We would also like to thank all of our individual Rare Disease Day donors, those who sent pictures of themselves featuring the ACDA logo, wore #JeansForGenes and those who participated in our #Reach1000Likes campaign!



We loved receiving all of the pictures of friends and family wearing items from the [ACDA Spreadshirt store](#) featuring the ACDA logo on Rare Disease Day! We received pictures of grandparents, rainbow babies, parents, other extended family members and friends! It was so inspiring to see the ACDA community come together and show off the logo. **Click [HERE](#) to visit the ACDA website and see a slideshow of our pictures and updates from Rare Disease Day!**

As part of our #Reach1000Likes campaign in connection with Rare Disease Day, our public ACDA Facebook page went from approximately 750 Likes to over 1,000 Likes in less than twenty-four hours!

Please click [HERE](#) to Like our page and continue to spread awareness!



Newspaper Articles (Pennsylvania, USA and Missouri, USA):

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

- **In memory of Callie Renee Murray**
(May 15, 2015 – May 23, 2015)
[Family Hopes to Raise Awareness of Rare Disease](#)
The Leader-Vindicator, February 24, 2016
Mercer, Pennsylvania, USA
- **In memory of Finley Kathleen Smith**
(March 18, 2015 – April 14, 2015)
[Couple Raises Awareness of Rare Diseases](#)
Lee's Summit Journal, February 26, 2016
Lee's Summit, Missouri, USA

Please also read the following article about an ACDA family in connection with their daughter's successful lung transplant in 2012:

- **Ellie Scott**
[Paying it forward: Family of double lung transplant recipient ready to give back through new foundation](#)
Lake News Online, January 26, 2016
Camdenton, Missouri, USA

Proclamation (North Carolina, USA):

Please click [HERE](#) to view a Proclamation issued by the State of North Carolina under official seal declaring February 20, 2016 as "Alveolar Capillary Dysplasia (ACD) Awareness Day" in the State of North Carolina, USA in honor of Johnny Rista (February 20, 2013 – March 4, 2013).

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 Spreadshirt:

The accrued commission payments from Spreadshirt between December 15, 2016 and April 13, 2016 is **\$46.75**. Items with the ACDA logo are available for purchase in our Spreadshirt store [HERE](#).

Update from AmazonSmile:

The ACDA was issued a **\$99.59** donation from the AmazonSmile Foundation as a result of AmazonSmile program activity between October 1, 2015 and December 1, 2015. 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>

Other Fundraising News:

If anyone knows of a printer that would donate the printing of our letterhead and brochures, please contact us at secretary@acdassociation.org.

Update from NORD:

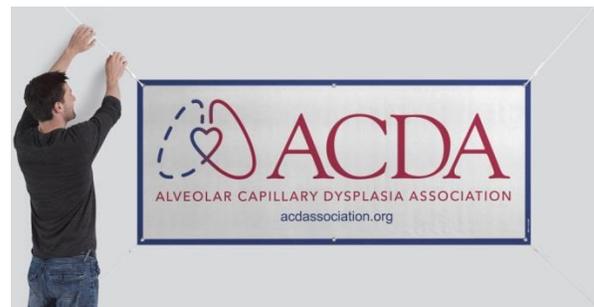
The NORD balance of the Research Fund (ACD) will be over **\$53,500.00** as of May 1, 2016.

The ACDA and The David Ashwell Foundation are pleased to announce the above balance exceeds the required minimum amount (\$33,500) for the issuance of a 2016 grant! NORD is in the process of revising the schedule of the grant cycle and has indicated the prior annual deadline of March 1 will now be revised to an early May deadline beginning with the current 2016 grant cycle. We expect NORD will announce the Request for Proposals (RFPs) by June 2016, which is anticipated to set forth a \$50,000.00 grant to be issued in 2016 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.

Let's start work on the 2017 grant!

Banner:



The ACDA is excited to announce the addition of two new custom ACDA banners! Both banners are 2.5' x 6' and are



made of durable, waterproof vinyl with grommets for hanging. The intended use of the banners is to allow ACDA registered families to request use of a banner for their fundraising and awareness events!

If you would like to request use of a banner in the U.S., please send an email to secretary@acdassociation.org to discuss sign-up for availability and shipping information. Availability will be on a first come, first serve basis and the requesting family is responsible for shipping costs to the next location on the sign-up sheet or return to the Secretary. Poster tubes for shipping can be obtained for free at U.S. Postal Offices. Shipping costs will vary but an example cost was \$11 to ship a banner from North Carolina to Missouri.

Please feel free to request the use of a banner if you have an event coming up! We hope the use of a banner will help our ACDA registered families in the United States coordinate and organize their own fundraisers and awareness events for ACDMPV research!

In honor of Finley Smith:

On March 18th and 19th, 2016, to celebrate their daughter's first birthday, Stephanie and Freddie Smith hosted the **1st Annual Finley Kathleen Memorial Wiffleball Tournament** (complete with a Home Run Derby!). And what a celebration it was!!! The tournament consisted of 13 teams playing on two separate fields at Finley Park (Freddie and Stephanie's very large backyard).

It was a truly inspirational day as over 100 people attended an event that had to deal with typical March weather in Missouri (strong winds with temperatures in the 30's and 40's). No one cared, though, as they were kept warm from not only the competition and a fire pit but also from the love on display throughout the day.

One of the most special moments of the entire weekend's celebration was when **Shannon and Shawn Cone of Olathe, KS whose beautiful daughter Kaylin Marie passed away from ACDMPV in 2000**, arrived to participate and the two families met for the first time.

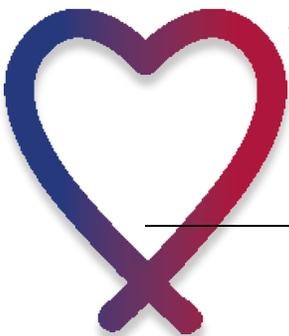
While there were a lot of home runs hit and great plays made in the field, the star of the day was most definitely Finley Kathleen. For on a day spent celebrating her first birthday, it was Finley who gave the greatest gifts of all. She brought together so many people who shared in an experience none will soon forget. **And these people opened their hearts to donate over \$5,000 to ACDA!!! Way to go Finny!!!**



1st Annual Finley Kathleen Memorial Wiffleball Tournament! Hosted by the Smith Family and joined by the Cone Family

In honor of Jasmijn Boesberg:

Roelina Jut, mother of Jasmijn Boesberg (February 8, 2013 - February 23, 2013), received a donation of **€500** for The David Ashwell Foundation through her employer, Georg Fischer N.V. Each year her company donates money to four organizations and the employees make suggestions which organization it should be. Roelina made a request for one of the donations to be made



REPORT FROM THE DAVID ASHWELL FOUNDATION:

Thank you everyone for your contribution to our fundraising efforts.

It has been a busy few months. In the autumn I decided to hold a fundraiser to commemorate David's 5th birthday (and my 40th!). I booked a local venue and following the arrival of our second rainbow baby Faye, I slowly started to get the ball rolling... It was a lot of work, and in the weeks leading up to the event, which was a ceilidh, I repeatedly told myself how crazy this was with a 3-4 month old baby! It was crazy, but it was worth it. Everyone, including me, had a fantastic night and we raised over £5000 (\$7059) for ACDMPV Research in memory of David. This included £4000 (\$5647) we raised on the night and £1000 (\$1411) in matched funding from Barclay's bank (who encourage staff to get involved and then match the funding!).

A big thank you to everyone who has contributed to The David Ashwell Foundation's fundraising. Especially to Sarah Marriot who has been tirelessly fundraising and awareness raising since Seb's passing a year ago.

The other big news is that on 22nd February we transferred £18,257.23 to NORD for the ACD Research account.

Remember that 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>

Please raise awareness of our small charity via social media including Twitter and Facebook.

If anyone in UK would like to support us, or join the team please get in touch.

UK families, please don't forget to use The Giving Machine when you online shop! Using this has raised significant sums and in Germany the equivalent website has raised 260 Euros (\$285 thanks to Karin Titze-Frech, see the report in the last newsletter!). If any other European Country has a Giving Machine equivalent and a UK charity can be your charity, please get in touch!

Stamp donations continue to come through! Thank you all for your contributions. Please all collect your used stamps for me – even if you are in US, Australia anywhere in the world – please collect them and we will figure out a way of getting them here!

All UK & Europe based families are welcome to use The David Ashwell Foundation as a means of fundraising for ACDMPV Research.

For additional information, please contact Simon and Amelia.

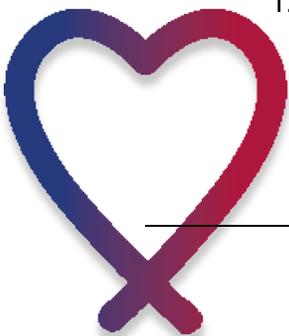
Website: <http://David Ashwell Foundation>

Email: davidashwellfoundation@yahoo.co.uk

Twitter: [@TDavidAshwellF](#)

Mobile (Amelia): 07855473686

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.





REMEMBERING OUR BABIES

[Redacted text block containing names and details of babies remembered]

[Redacted text block]

WELCOME TO NEW FAMILIES

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

- [Redacted family name]
- [Redacted family name]
- [Redacted family name]
- [Redacted family name]



- [REDACTED]
- [REDACTED]

- **Stephanie Smith** - Stephanie created the [Bravelets](#) fundraiser, implemented the “\$5 to Wear Jeans to Work” with her employer on Rare Disease Day and organized an individual fundraiser in March 2016 that raised significant funds for ACDMPV research.

The new member of the Communications Committee is:

- **Candice McCasland** - Candice has been an active Facebook participant and has a way of connecting with people. She is a great asset on the Communications Committee.

A big Thank You! to the new committee members and to the existing members for their continued contributions. The composition of the committees is now as shown below. If you have any ideas or suggestions, please always feel free to contact one of the committee members!

NEW ACDA COMMITTEE MEMBERS

Four additional ACDA committee members were approved in Special ACDA Board of Directors meetings in January 2016. All are great fits for their roles on their respective Committees.

Three new parents joined the Fundraising Committee and all bring experience from prior fundraisers for ACDMPV research:

- **Renee Murray** - Renee coordinated the [Giving Bean](#) fundraiser, implemented the “\$5 to Wear Jeans to Work” with her employer on Rare Disease Day and is in the process of organizing an individual fundraiser in May 2016 for ACDMPV research.
- **Kristen Rilling** - Kristen has coordinated an individual fundraiser since 2013 that has raised significant funds for ACDMPV research with over 500 donors. She also suggested the “\$5 to Wear Jeans to Work” campaign and implemented the event with her employer on Rare Disease Day.

Communications Committee:

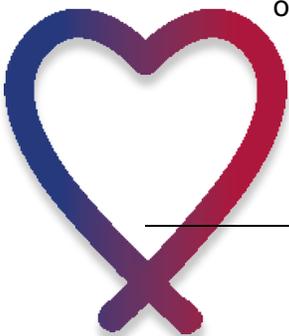
- Emily Eschweiler
- Diana Woida Locke
- Candice McCasland
- Eliza Rista

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>



SAFE ARRIVALS!

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

[REDACTED]

[REDACTED]

- [REDACTED]

[REDACTED]

- [REDACTED]

[REDACTED]

- [REDACTED]

CONNECT WITH US

Facebook:

- [Official ACDA Public Page](#)
- [Parent Group](#) (private)
- [Family Group](#) (private)

Read about the private groups on our website with information on how to join:

<http://acdassociation.org/support-groups/>

Twitter:

- Follow us [@acdassociation](#)

Website:

acdassociation.org

Email:

President@acdassociation.org (Steve Hanson)

Secretary@acdassociation.org (Eliza Rista)

Treasurer@acdassociation.org (John Rista)

A note from the Secretary: 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 Secretary@acdassociation.org.

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

