

# ACDA NOTES

FROM THE ALVEOLAR CAPILLARY DYSPLASIA ASSOCIATION

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## 2018: Year in Review

As we take a look back at 2018:

- Fourteen new families registered with the ACDA. These families are from the USA, France, Australia, Germany, Belgium, Sweden and Brazil.
- One lung transplant in Germany.
- One NORD research grant in the amount of \$50,000.
- Fifteen rainbow babies born to ACDA registered families.
- One participant in the British Transplant Games 2018.
- Two additional tattoos of the ACDA logo.
- More than ten 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

## AWARENESS NEWS

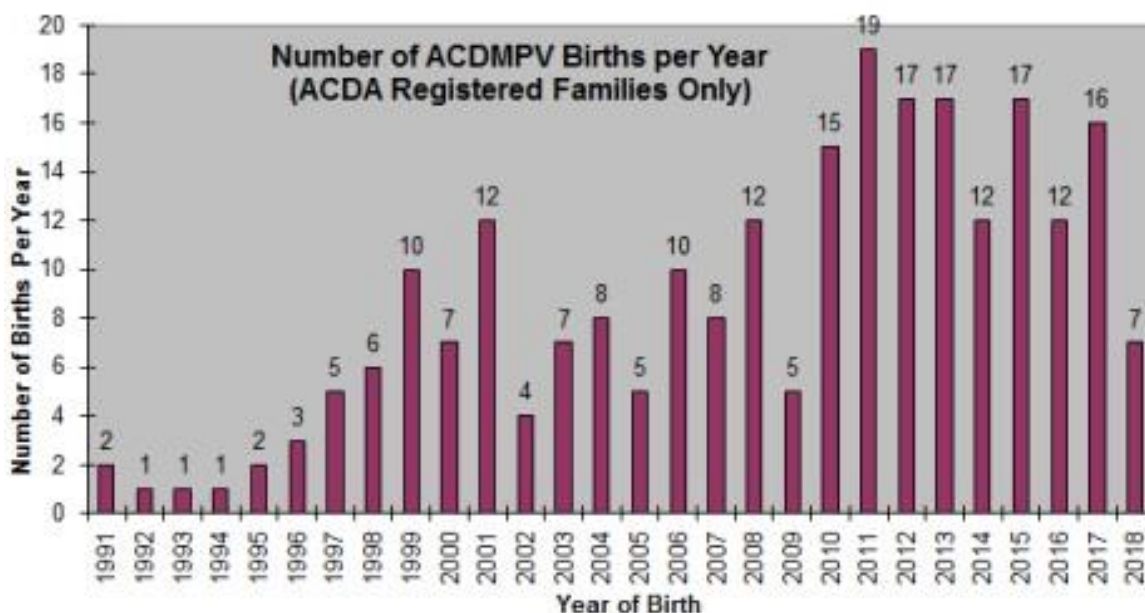
### Statistical Update (January 2019):

It has been about three years since the ACDA provided a statistical look at the ACDA community of registered families. *Please note the following data is **only** with respect to families that have registered with the ACDA and DOES NOT include all reported or known cases of ACDMPV. In addition, the known case reports almost certainly underestimates the true prevalence of ACDMPV.*

The ACDA hopes the charts and reports answer some of your questions and provide insight into ACDMPV. A couple of notes on the data:

- We are respectful of the privacy of families registered with the ACDA, and if you have requested that certain information not be shared, we suppress it in any data shared with others (e.g. the ACDA membership directory). However, all of the data is used to generate reports and charts because such information is not uniquely identifiable to a specific family.
- The data is intended to be accurate but please know some information may not be current or precise as the ACDA has lost contact with certain families over the years. Please always consult with your qualified medical professionals.

**1. How many babies with ACDMPV have been reported to the ACDA?** As of January 1, 2019, there are **244 confirmed or suspected ACDMPV cases in 233 families registered with the ACDA.** Note there is sometimes a delay by families in their registration with the ACDA, which means births in the most recent year may not have registered with the ACDA as of this statistical update.



(continued on following pages)



- 2. What are the chances that our next child will have ACDMPV?** This is the most common question submitted to the ACDA and it is a difficult question to answer as all of the cause(s) of ACDMPV are not known. This data is based on **the 233 families in the ACDA database:**

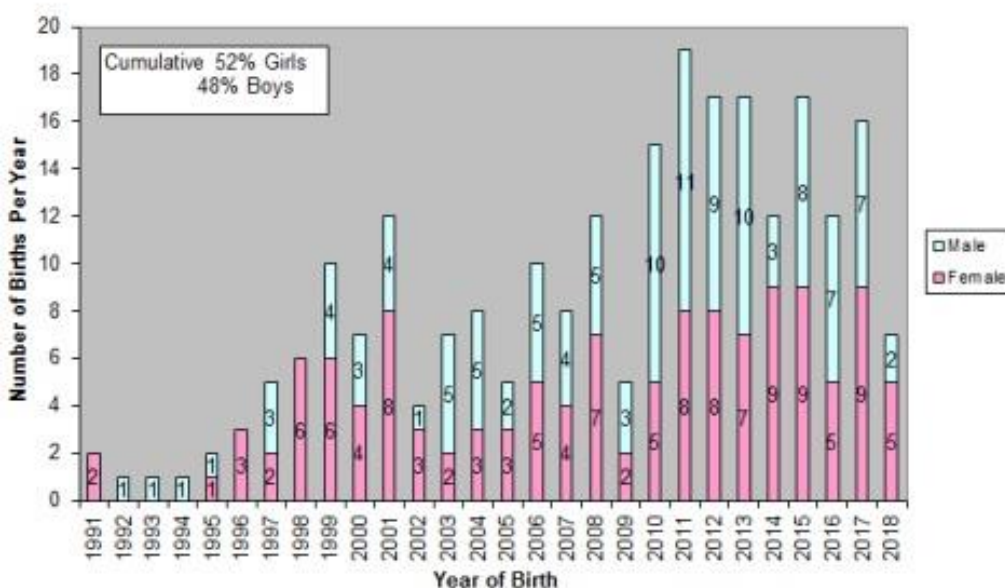
Families with one child born with ACDMPV:	224
Families with two children born with ACDMPV:	7
Families with three children born with ACDMPV:	2
Families that had a healthy child after an ACDMPV baby:	135
Families who have not had another child (or not known to us):	98

\* A Family is defined as children from the same parents (i.e. re-marrying would be another family).

\*\* Note recurrence numbers do not contemplate a small number of families registered with the ACDA that did not pursue a subsequent pregnancy after genetic testing from the initial occurrence of ACDMPV revealed a familial connection.

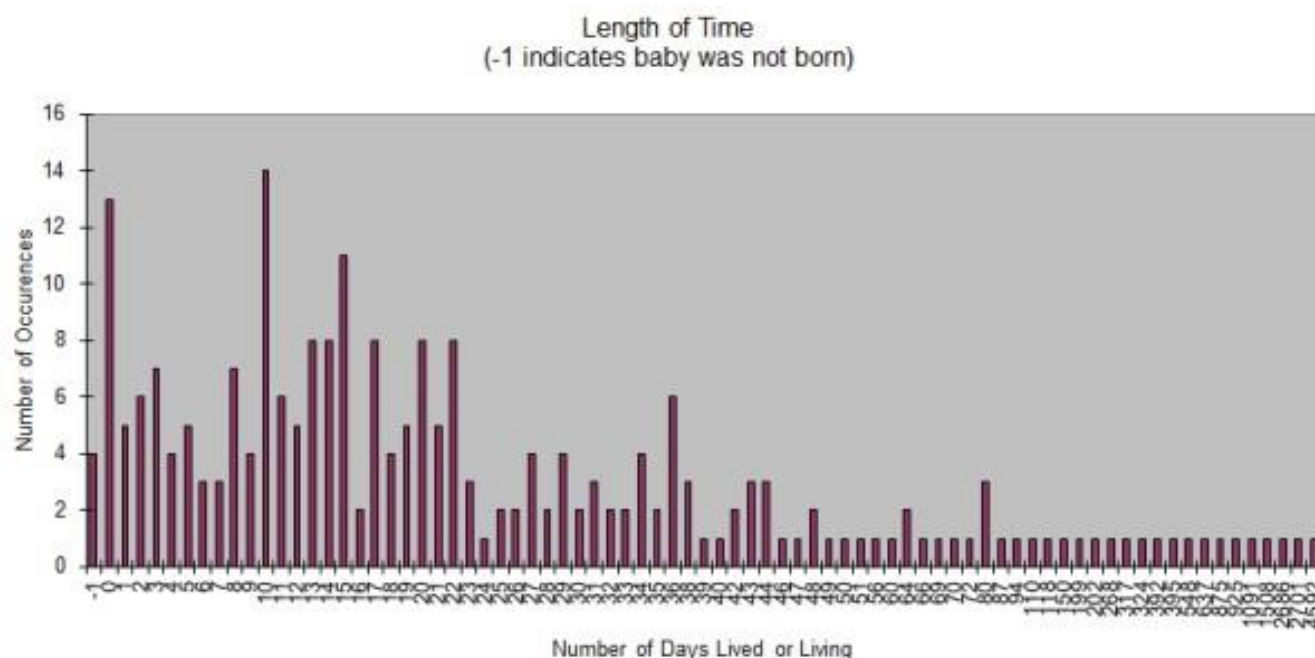
- 3. Does ACDMPV affect males and females equally?** The research papers we have seen indicate that ACDMPV affects males and females equally. As the sample size has grown over the years, the data is converging on the 50/50 ratio. The 2010 data was 57% female and 43% male, 2012 showed 52% female and 48% male and 2015 showed 51% female and 49% male. Current data in January 2019 shows 52% female and 48% male.

**Number of ACDMPV Births per Year  
and Number of Males and Females  
(ACDA Database Only)**



4. **How long do babies with ACDMPV live?** ACDMPV is considered a fatal diagnosis, however, some patients have survived with atypical or late presenting ACDMPV long enough to receive lung transplants. Lung transplantation is available in limited circumstances and there is otherwise no known cure, supportive therapy or treatment for ACDMPV.

ACDMPV often presents itself within a few hours or days while the baby is still in the hospital, but in several cases it has not presented until weeks or months later. When the symptoms present and the level of care available to an ACDMPV baby affect how long the baby will live. The ACDA does not have specific data on when babies presented with ACDMPV or what levels of care were provided, but the following chart shows that children live from less than a day to more than a year, in limited cases.



\*Note, the chart above includes seven children living with a presumptive or confirmed ACDMPV diagnosis who received a successful bilateral lung transplant (BLT) or living without a BLT (as of January 1, 2019). See question #5 below for further information about these specific cases.

5. **How many children registered with the ACDA have received lung transplants or are living with an ACDMPV diagnosis without lung transplant?** This data is based **only** on the 233 families *in the ACDA database*:

Bilateral lung transplant recipients*	5 (4 successful outcomes)
Living with a presumptive or confirmed ACDMPV diagnosis <i>without</i> a bilateral lung transplant	3

\* Note there are several other successful lung transplants for ACDMPV patients discussed in medical research journal articles but not all recipient families have contacted the ACDA.



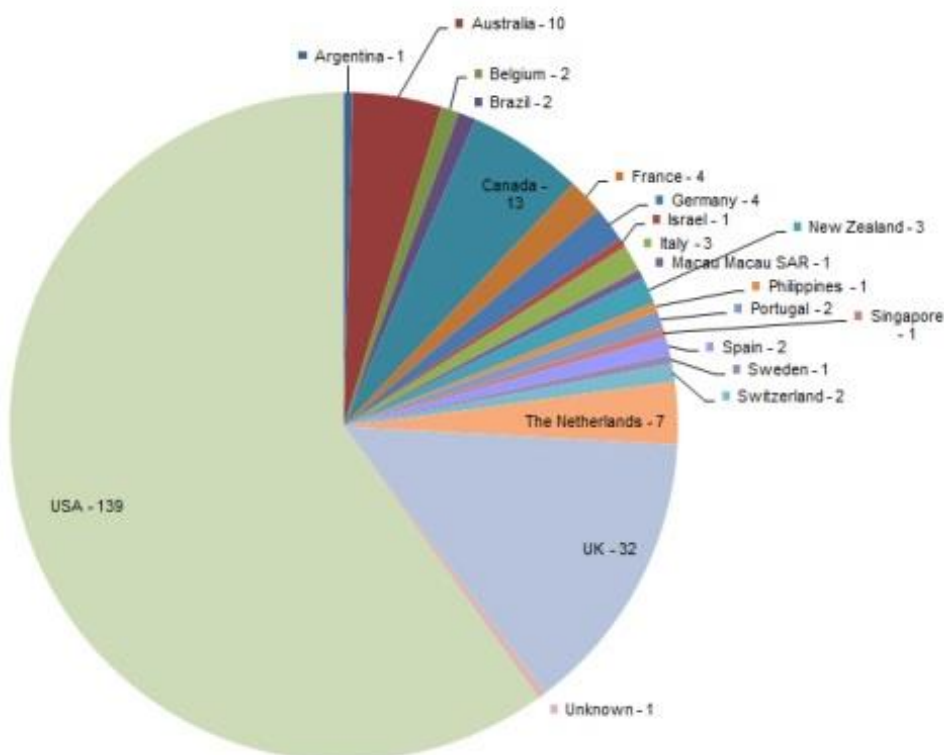


**6. How many families registered with the ACDA have received a presumptive prenatal diagnosis of ACDMPV based on prenatal genetic testing without a familial history of ACDMPV?** This data is based **only** on the 233 families *in the ACDA database*:

Presumptive prenatal diagnosis without familial history of ACDMPV 5

**7. Where do ACDA families live?** ACDA families are currently located in 21 countries throughout the world. The largest groups remain the same since the last chart with approximately 60% in the USA, 14% in the UK and 6% in Canada.

Number of ACDMPV Cases per Country - ACDA Members Only



**8. Are there any ACDMPV affected families that live near me?** View [Registered ACDA Family Locations](#) in a full screen map. This map sets forth the location of a majority of ACDA registered families, as sorted in chronological order by birth date of their child(s) affected by ACDMPV. Markers show the approximate location via city and state without disclosing specific street addresses, names or any other personally identifiable information.

\*Note, this map will not exactly match other statistical information provided by the ACDA as not all members are represented on the map if privacy settings during initial registration restricted the sharing of their location.

\*\*In addition, there are a handful of families included on the map whose ACDMPV initial diagnosis was later retracted by their medical professionals. Note these families with a retracted diagnosis are \*not\* included in ACDMPV statistical information provided by the ACDA but their location is included on the map and in our membership directory as the loss of a child is a common bond and connections or outreach may be beneficial, regardless of the final diagnosis.





### Families in the News (Dean Grant):

Derek Grant, ACDA father to Dean (September 13, 2016 - July 27, 2017), recently created a video called “Dean’s Story: A Happy Baby through the Most Difficult of Times,” which was published by Media Milwaukee in December 2018. Please click [HERE](#) to watch the inspiring video about Dean (see also Issue #60 of ACDA Notes).



Dean and Ashley Grant share a video story about their son, Dean

### Día de los Inocentes (Billie Paras):

Genesis Paras, ACDA mom to Billie (April 14, 2018 - April 16, 2018), invited ACDA families to be a part of her family’s observances for *Día de los Inocentes* (Day of the Innocents) on November 1, 2018, which is part of the *Día de los Muertos* multi-day holiday that gathers family and friends to remember those who have passed away. The Paras family invited ACDA families to send a picture of their child if they would like to be included in the Paras’ memorial alter, decorated with orange marigolds, candles and a big piece of pan de muerto. The families who participated received a pressed marigold flower, a custom candle to light next year, and a picture of the Mariachi singing to the alter. The Paras family also shared the experience live on Facebook. Thank you to the Paras family for their thoughtfulness.

## RESEARCH NEWS

### Journal Article (Am J Human Genetics):



The genetic research team at Baylor College of Medicine in Houston, Texas, USA recently collaborated with an international team to publish a manuscript entitled “Complex compound inheritance of lethal lung developmental disorders due to disruption of the *TBX-FGF* pathway” in the American Journal of Human Genetics, which can be found [HERE](#). The primary defects in lung branching morphogenesis, resulting in neonatal lethal pulmonary hypoplasias, are incompletely understood. This paper examined the pathogenetics of human lung development by studying a unique collection of patient samples with several different kinds of clinically and histopathologically diagnosed interstitial neonatal lung disorders: acinar dysplasia (AD) (n = 14), congenital alveolar dysplasia (CAD) (n = 2), and other rare lethal lung hypoplasias (n = 10). **Note patients with ACDMPV were not included in this study but AD, CAD and other lung hypoplasias are similar rare lethal lung pulmonary diseases.**

The research team identified rare heterozygous copy-number variant deletions or single-nucleotide variants (SNVs) involving *TBX4* or *FGF10* in greater than 60% of the studied individuals. In addition to *TBX4*, overlapping deletions at 17q23.1q23.2 identified in seven individuals with lung hypoplasia also remove a lung-specific enhancer region. Individuals with coding variants involving either *TBX4* or *FGF10* also harbored at least one non-coding SNV in the predicted lung-specific enhancer region, which was absent in 13 control individuals with the overlapping deletions but without any structural lung anomalies. The occurrence of





rare coding variants involving TBX4 or FGF10 with the putative hypomorphic non-coding SNVs implies a complex compound inheritance of these pulmonary hypoplasias. Moreover, they support the importance of TBX4-FGF10-FGFR2 epithelial-mesenchymal signaling in human lung organogenesis and help to explain the histopathological continuum observed in these rare lethal developmental disorders of the lung.

## LUNG TRANSPLANT UPDATES

### *Lung Transplant Update (Tom Schnierle):*

The ACDA introduced you to Tom in July 2018 as an almost two year old under the in-patient care of a German hospital living with a presumptive ACDMPV diagnosis based on genetic testing and clinical symptoms, but



**Tom, a two year old ACDMPV lung transplant recipient in Germany, a few weeks post-transplant in January 2019**

without a bilateral lung transplant (see Issue #64 of ACDA Notes). At the time, Tom had been in the intensive care unit **since birth** and a lung biopsy was pending confirmation at Baylor College of Medicine (biopsy subsequently did confirm ACDMPV in January 2019). At the time of our last update, his family's goal was

to finally bring Tom home with nitric oxide and the family was also considering a possible bilateral lung transplantation pending discussions and review with Tom's

medical professionals, which was complicated by limited organ availability and the fact that lung transplants were not performed on small children in Germany.

**We are pleased to report Tom was listed for transplant in October 2018 and received a bilateral lung transplant at the end of December 2018 in Germany.** A few weeks after the successful transplant, Tom's mother, Stefanie, shared the first pictures of Tom without any oxygen support for the first time in his 2+ years.

Our heartfelt congratulations to Tom and his family! #acdwarriors

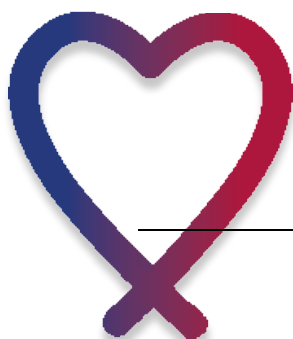
### *Lung Transplant Update (Luca Palmisano):*

We wrote about Luca Palmisano in Issues #57, #58, #59 and #61 of ACDA Notes and he continues to make the news! Luca was recently invited to Old City Hall for a private showing of the city's annual Holiday Train Show. Click [HERE](#) to read the touching story in The Burlington County Times.

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



**Luca, a lung transplant recipient, enjoys a private showing of a holiday train show**





**Donations:**

[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.

The minimum amount required for the issuance of a 2019 NORD grant is \$35,000.

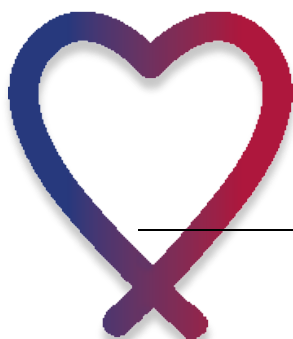
The balance of the ACDA bank accounts as of January 31, 2019 is \$ [REDACTED].

The ACDA was issued a \$[REDACTED] donation from the AmazonSmile Foundation as a result of AmazonSmile program activity between July 1 and September 30, 2018. 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>

The accrued commission payment from Spreadshirt

**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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[illegible]

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



## #GivingTuesday Update:



**WOW.** The ACDA's fundraising goal for #GivingTuesday on November 27, 2018 was \$1,000 and, with your help, we were able to bring in just over **\$2,630!!**

**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.

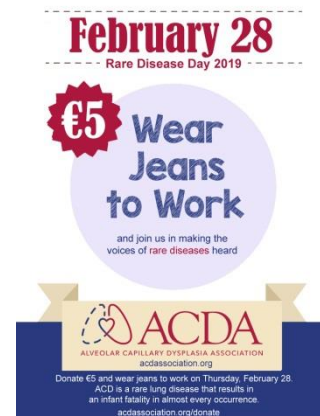
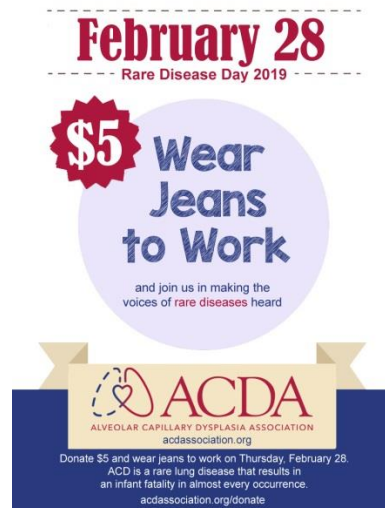
Thanks very much for the generous #GivingTuesday donations in honor of *Kinsley Labutka, Finley Smith, Fraser Dunne, Ayla Prushansky, Fallon Rilling, Amelia Weaver, Erik Lund, Dean Grant and Sadie McCasland, Marleigh Sanders, Ainsley Abodeely, Annabelle Logue, Ronan Bush, Phoebe Bush, Eric Hanson and Johnny Rista.*

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 2018.

## Wear Jeans to Work Flyers:

As referenced on page 6, 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 Thursday, 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](http://acdassociation.org/donate) and we are happy to track the amounts donated in your child's honor.





### **Birthday Fundraiser (Aurora Splawn):**

In August 2018, Tiffany Hensley, ACDA mom to Aurora (July 23, 2016 - August 6, 2016), organized a birthday fundraiser for **ACDMPV research in honor of Aurora**. In Tiffany's own words, "For my birthday this year, I'm asking for donations to the Alveolar Capillary Dysplasia Association. I've chosen this nonprofit because their mission means a lot to me, and I hope you'll consider contributing as a way to celebrate with me. Every little bit will help me reach my goal. If I get nothing else for my birthday but a small donation in honor of our beautiful daughter I would be forever grateful. Finding a cure will only be possible through extensive research which is funded solely on donations. Let's help find a cure for any future ACD warriors. "SEARCHING FOR ANSWERS, HOPING FOR A CURE"



**Tiffany exceeded her \$100 goal by raising \$105 through a Facebook birthday fundraiser in honor of her daughter, Aurora Splawn. Thank you Tiffany!!**

### **School Fundraiser (Imogen Bolton):**



In December 2018, **Aliessia, age 13**, 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, cakes and other bits and bobs. Aliessia also displayed the massive poster she created in the past explaining all about ACDMPV and Imogen's story. **She raised £76.51 (\$100)!!** The ACDA is so incredibly proud of this young lady's hard work and dedication to ACDMPV research!

### **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>



## REPORT FROM STICHTING ACD (NETHERLANDS):



Mathijs Lourens, ACDA father to Myla (November 20, 2015 - December 17, 2015), continues his amazing work in support of his Dutch ACDMPV foundation, "[Stichting ACD](#)." Mathijs plans to continue running marathons for ACDMPV awareness in Europe and then worldwide! He previously completed the Rotterdam Marathon (April 2017 and April 2018), the Amsterdam Marathon (October 2016 (half), October 2017 and October 2018 (Kids Run)), and various other running events. **Please contact Mathijs if you are interested in participating or cheering on his upcoming plans to raise ACDMPV awareness in the Netherlands, Germany or beyond!** For additional information about the upcoming runs highlighted below, please visit [Run4ACD](#) on Stichting ACD's website and [Groet uit School! Run](#), [NN Marathon Rotterdam](#) and [BMW Berlin Marathon](#).

### **TCS Amsterdam Marathon Kids Run (October 21, 2018):**



Mathijs and **kids** from team Stichting ACD ran the **TCS Amsterdam Marathon Kids Run on October 21, 2018** to raise ACDMPV awareness! The Kids Run is a children's run that takes place during the TCS Amsterdam Marathon. Children aged 4 to 12 can participate in the 700-metre



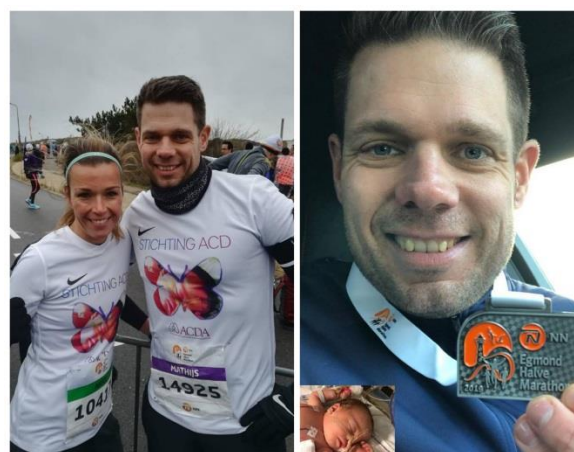
run, which starts behind the stadium and finishes in the stadium itself. A particularly proud moment for Mathijs was watching **his young son, Alec, participate in the event in honor of Alec's sister, Myla.**



### **NN Egmond Halve Marathon (January 13, 2019):**



Mathijs and team Stichting ACD also ran the **NN Egmond Halve Marathon on January 13, 2019** to raise ACDMPV awareness!





### **Groet uit Schoorl Run (February 10, 2019):**



Upcoming Event: Mathijs will be running the **Groet uit Schoorl Run in North Holland on February 10, 2019**. The Groet uit Schoorl Run attracts 9,000 runners to this North Holland resort and has been held every year on the second Sunday of February since 1979.

### **NN Marathon Rotterdam (April 7, 2019):**



Upcoming Event: Mathijs and Stichting ACD will run the **Rotterdam marathon on April 7, 2019 for the third year in a row**. 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!

### **BMW Berlin Marathon (September 29, 2019):**



Upcoming Event: Chasing the dream to cross the world with Myla! Mathijs trained hard with focus and determination for many months in order to expand his marathon adventure across the border in October 2018 (see Issue #65 of ACDA Notes). For his first foreign marathon, he planned to run the marathon in Cologne, Germany on October 7, 2018 to raise ACDMPV awareness in Germany. Despite his best efforts and

tremendous optimism, a significant cold mandated that Mathijs sit out his first foreign marathon attempt. But Mathijs never gives up and is already planning his next marathon adventure in Germany! Mathijs plans to participate in the **BMW Berlin Marathon in Berlin, Germany on September 29, 2019!**

### **Bracelets for Myla's 3<sup>rd</sup> Birthday:**



Jessica Anderson, ACDA mom to Myla, and Mathijs worked on a cool collaboration for a fundraiser for Myla's 3<sup>rd</sup> birthday in November 2018. Bracelets specially made for Myla's birthday and featuring special mantras were sold for € 9,95 each with all proceeds going to Stichting ACD. *Breathe, Love, Believe, Embrace, Magic and Faith*. This is such a beautiful way to raise funds for ACDMPV research in honor of Myla!

### **Angel Gallery:**

You are invited to view the "[Angel Gallery](#)" on Stichting ACD's website for a touching image display featuring many of our ACDA babies. If you would like your child to be included in the gallery, please contact Mathijs at [contact@stichtingacd.nl](mailto:contact@stichtingacd.nl) and send him a picture with full name, birth date and angel date.





## REPORT FROM THE DAVID ASHWELL FOUNDATION (UK):

In May 2018 we transferred £16,221 (\$21,332) to the ACDMPV research fund at NORD. Since March 2011 **£201,737 (\$263,409)** 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 are always very enthusiastic to raise more money for ACDMPV research and would appreciate any help! **Thank you to those families who have set up direct debits for the charity and whose families fundraise at work for us.**

Thank you to those who have set up Rare Disease Day fundraisers at their schools and workplaces.



**At the moment not many people are using The Giving Machine.**

Please, UK families can you use The Giving Machine ([GO HERE](#)) 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! Watch [HERE](#) how it works! I have the giving machine app on my phone and it makes giving as I shop so easy. It's very quick and easy to [sign up](#)!



Please use **The David Ashwell Foundation Virgin Money Giving pages**. On our Virgin Money Giving pages, **set up a page in for your precious baby/ child and encourage people to donate there.**

We are here to help UK families fundraise for ACD research so that we can help to find answers to the condition that has become such a major part of our lives.

Simon and I run this as well as working and bringing up our children, Auden and Faye. We are

very, very grateful to anyone who uses this 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).

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.
2. Fundraise while you online shop ([The Giving Machine](#)) (a percentage of what you spend is donated)
3. Collect [postage stamps](#)



**The 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://DavidAshwellFoundation>

**Email:** [davidashwellfoundation@yahoo.co.uk](mailto:davidashwellfoundation@yahoo.co.uk)

**Twitter:** [@TDavidAshwellF](https://twitter.com/TDavidAshwellF)

**Mobile** (Amelia): 07855473686



## REMEMBERING OUR BABIES

[REDACTED]

[REDACTED]

[REDACTED]

[REDACTED]

## Welcome to New Families

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

- [REDACTED]
- [REDACTED]
- [REDACTED]



