

ACDA NOTES

From The Alveolar Capillary Dysplasia Association

Volume 17, Issue 2

<http://www.acd-association.com>

Spring 2015

Dear Families and Friends,

Spring is a time of rebirth and inspiration and a great time to envision new possibilities. It is also a time we all have renewed energy and hope. I hope that this spring is a time of transformation and happiness for you and your families.

At the ACDA, several transformations are underway. A new logo, new website and a new look for the newsletter are among the items undergoing change. Look for those later this summer. In the meantime, enjoy reading about all that is going on in the ACD community.

Many thanks again to all the families and friends that raised money or donated to the ACDA or NORD that will enable us to fund a research grant for approximately \$55,000. NORD plans to initiate this grant process soon.

Fondly,

Steve Hanson
ACDA President



ACDA Board Meeting Held

The ACDA Board of Directors held its quarterly meeting on April 28, 2015 to discuss various topics. The following items were on the agenda:

- Reviewed the logo recommendations from the Communications Committee and voted to proceed with one design.
- Accepted a recommendation from the Fundraising Committee to undertake a coffee fundraiser.
- Discussed the progress of our website update being led by Eliza Rista.

Please feel free to contact any of the Board members with any comments or questions you have.

New Changes Coming to the ACDA

The ACDA has really big news!! We are so excited to announce a fantastic new logo has been selected and will be introduced this summer. In addition, the ACDA website is undergoing a complete site redesign, including new content and a mobile friendly format. We are also in the process of creating a terrific new storefront, a revamped newsletter layout and updated informational brochures. All of these changes and more are in the process of being finalized and will be unveiled simultaneously with the distribution of the next newsletter. We are so thrilled with the new look and direction of the ACDA!! Keep an eye out for the official unveiling later this summer!

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Research Updates

Summary of

Genomic and Epigenetic Complexity of the FOXF1 Locus in 16q24.1: Implications for Development and Disease By A V Dharmadhikari, P Szafranski, V Kalinichenko and P Stankiewicz, 2015

This is a review article, summarizing recent studies of FOXF1. It discusses where in the body the FOXF1 gene is expressed in humans and mice, the effects on mice who have the FOXF1 gene removed, the effects of duplication of FOXF1 and FOXF1 and cancer. Of relevance to ACD parents, the authors point out that in ACD-affected infants, 44 mutations involving FOXF1 and 36 deletions involving FOXF1 or upstream of FOXF1 have been reported thus far. In infants with ACD for whom it was possible to determine from which parent a deletion arose, in all 24 they arose *de novo* on the maternal chromosome, consistent with the understanding the FOXF1 is paternally imprinted in human lungs.

Thanks to Dr. Simon Ashwell, father to David, for this summary.

Raising Awareness

- John Rista (father of Johnny Rista, February 20 – March 4, 2013) - On March 25, 2015, the North Carolina Rare Disease Coalition hosted a Rare Disease Legislators' Breakfast at the state capitol in Raleigh, North Carolina. This event was an opportunity for state legislators to learn about current research and development opportunities within our state with respect to rare diseases and also to propose support for the establishment of a N.C. Rare Disease Advisory Council via legislation. The coordinator of the event, Taylor's Tale President Sharon King, who has a daughter battling a rare disease, emphasized how it is North Carolina's goal to be a leader in rare disease research and development. In addition, speeches were given by Jude Samulski, director of the Gene Therapy Center at UNC-Chapel Hill, and Jon Stonehouse, president and CEO of BioCryst Pharmaceuticals. Since this meeting, House Bill 823 was introduced into the N.C. House and passed unanimously. It is now in the N.C. Senate and will hopefully meet a quick unanimous passing vote there as well. I made a few connections on behalf of ACD and was grateful for the opportunity to attend this meeting devoted to raising the profile of rare diseases within my home state.
- Kristin Rilling (mom to Fallon, October 10-21, 2013) - Read about Kristen's fundraising for ACD in a NORD blog in honor of Mother's Day and Fallon: <http://blog.rarediseases.org/mothers-day-celebrating-amazing-moms-in-the-rare-disease-community/>
- Jennifer Hanson (little sister to Eric, June 7-17, 1997) – Jennifer had a 6th grade English assignment that required her to research a group or organization of her choice that has had an impact on the world. She chose the ACDA! She had to create a poem, a poster, and a PowerPoint presentation to present to her class. Jennifer is a great reminder that even our kids can help us raise awareness!

Jennifer and her project on the ACDA



Report from The David Ashwell Foundation

Our balance as of 28th May 2015 was £4874 (\$7,626).

Since March 2011, we have raised **£142,282 (\$222,620)** for ACD Research. The David Ashwell Foundation has contributed to three recent grants for ACD research issued by NORD: £33,118 (\$52,053) in February 2012, over £95,011 (\$157,072) in March 2014 and £9278 in March 2015. All the money raised is from ACD families and friends in UK and Europe.

Recent fundraisers include Simon Ashwell (David's Dad) running the Edinburgh marathon with a family friend, Dariush Kamali. Simon and Amelia did a number of newspaper interviews and Simon did a live radio interview with our local station BBC Radio Tees.

<http://www.gazettelive.co.uk/news/teesside-news/stokesley-dad-remember-baby-son-8997643>

To date this fundraiser has raised over £4,220.25 (\$6,603). Simon ran the marathon in 3:14 and Dariush in 3:45 (despite knee injuries!). Amelia, Auden and Simon's sister Gemma were there to support the runners. (See pictures page 4)

Another Ashwell family friend, Mike Charlton, recently completed a rather unique challenge where he was fundraising for a number of charities including ours. This challenge was to tackle the five highest peaks in Ireland, Northern Ireland, Scotland, England and Wales by bike and hike. Mike and the team of peaky bikers set off on 7th June scaling the peaks and cycling the 960 miles between them in only 7 days!

They completed:

960 miles of cycling

50 miles of walking

50,506 feet of climbing either on the bike or on foot

There was approximately 90 hours on the bike and 25 hours of walking and not much sleeping. It was an amazing challenge and had us all gripped on Twitter, Facebook and the occasional phone call where Mike sounded more and more tired!

Thank you to all the ACD families who sent messages of support and encouragement to Mike throughout the week-long challenge. I know he appreciated it!

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 shop! Using this has raised £754 (\$1,180) since 2011 and in Germany the equivalent website (thanks to Karin!). 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 collect all 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 ACD Research. If you live in the UK (and elsewhere), there are a number of options available for funding ACD 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 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>

For additional information, please contact Simon and Amelia. <http://David Ashwell Foundation>
davidashwellfoundation@yahoo.co.uk

Twitter: @TDavidAshwellF

Amelia's mobile: 07855473686

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



Simon and Dariush pre-marathon in chilly Edinburgh

Dariush and Simon with their medals just having completed the marathon



From the Fundraising Committee

The Fundraising Committee held a meeting on April 17, 2015. The following items were discussed:

- The committee voted to recommend a coffee fundraiser to the Board of Directors.
- A replacement for Café Press as our on-line vendor was discussed with further research required.
- In January 2015, the IRS notified the ACDA that the organization had been selected for an audit for the year 2013. The ACDA was randomly selected from a group of non-profit organizations that had received quick approval from the IRS in 2013. The ACDA supplied a detailed description of activities during 2013 as well a statement of revenues and expenses. The IRS auditor recently notified the ACDA that there were no findings to dispute our non-profit status, and we are in good standing with the IRS.

John Rista and Donna Hanson
Fundraising Committee Members

Remember when you shop on line, support the ACDA by shopping on Amazon Smiles. They make a donation to the ACDA for every purchase made.



From the Communications Committee

The ACDA Communications Committee has undertaken quarterly meetings since January 2015 and has subsequently agreed to increase the frequency of official meetings to convene the first Monday of each month pending the availability of matters to address. If there are any suggestions or ideas you would like the Communications Committee to discuss, please feel free to reach out to ACDA Communications Committee members Kim Anderson, Emily Eschweiler, Diana Locke or Eliza Rista at any time - we would love to hear from you!

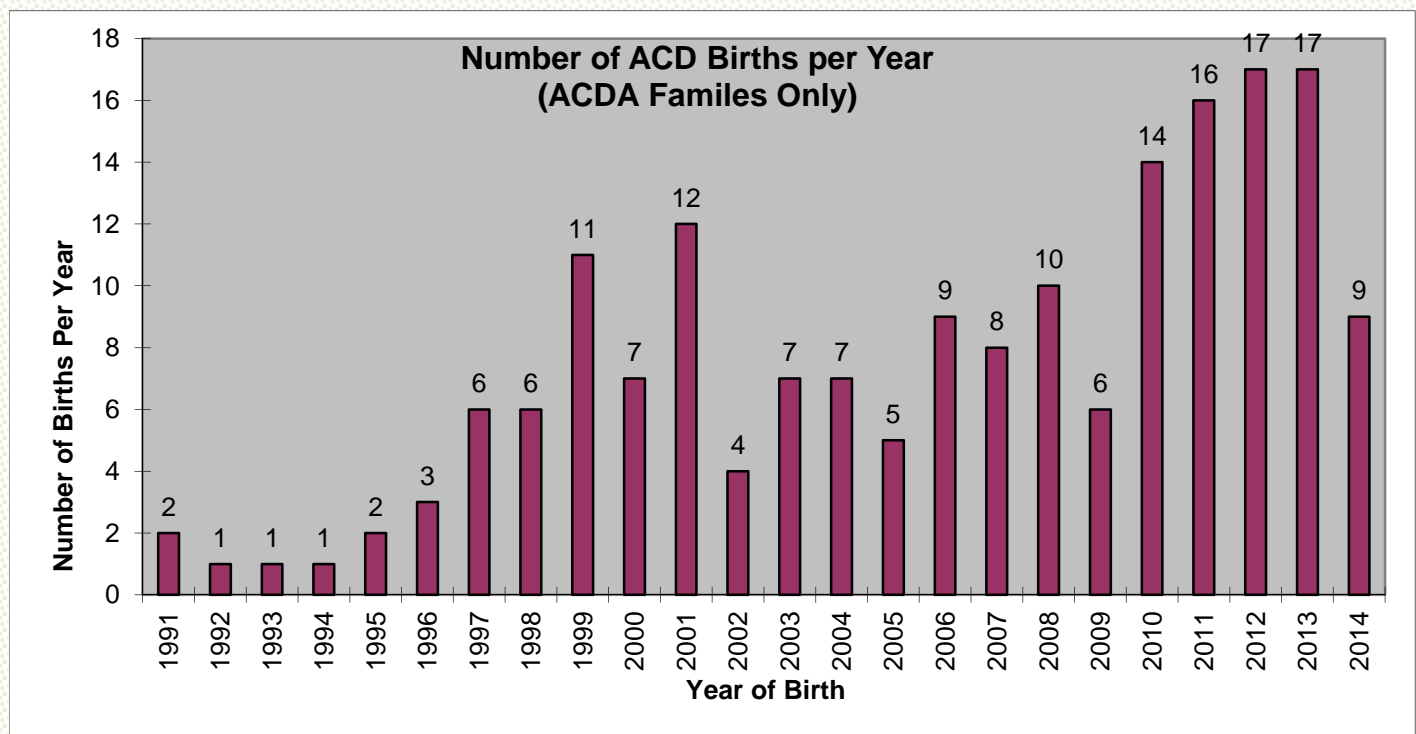
ACDA - Who Are We?

It has been about three years since we provided a statistical look at the ACDA community. The following **data is only on families that have registered with the ACDA and DOES NOT include all reported or known cases of ACD**. We hope the charts and reports answer some of your questions and provide insight into ACD.

A couple of notes on the data:

- a. We are respectful of members' privacy, and if you have requested that certain information not be shared, we suppress it in any data shared with other people (e.g. the ACDA Directory). However, all of the data is used to generate reports and charts because that information is not uniquely identifiable to a specific family.
- b. The data is accurate to the best of our knowledge, but we know that some is not current as we have lost contact with several families. However, we believe it is reasonably representative of the ACDA families that have registered.

1. How many babies with ACD have been reported to the ACDA? Since the 2012 update, there have been an additional 42 cases bringing the total to 177 live births and 4 unborn. The increasing number of ACD children starting in 2010 could indicate that knowledge and awareness of ACD in the medical community is on the rise and not necessarily that there are more cases.



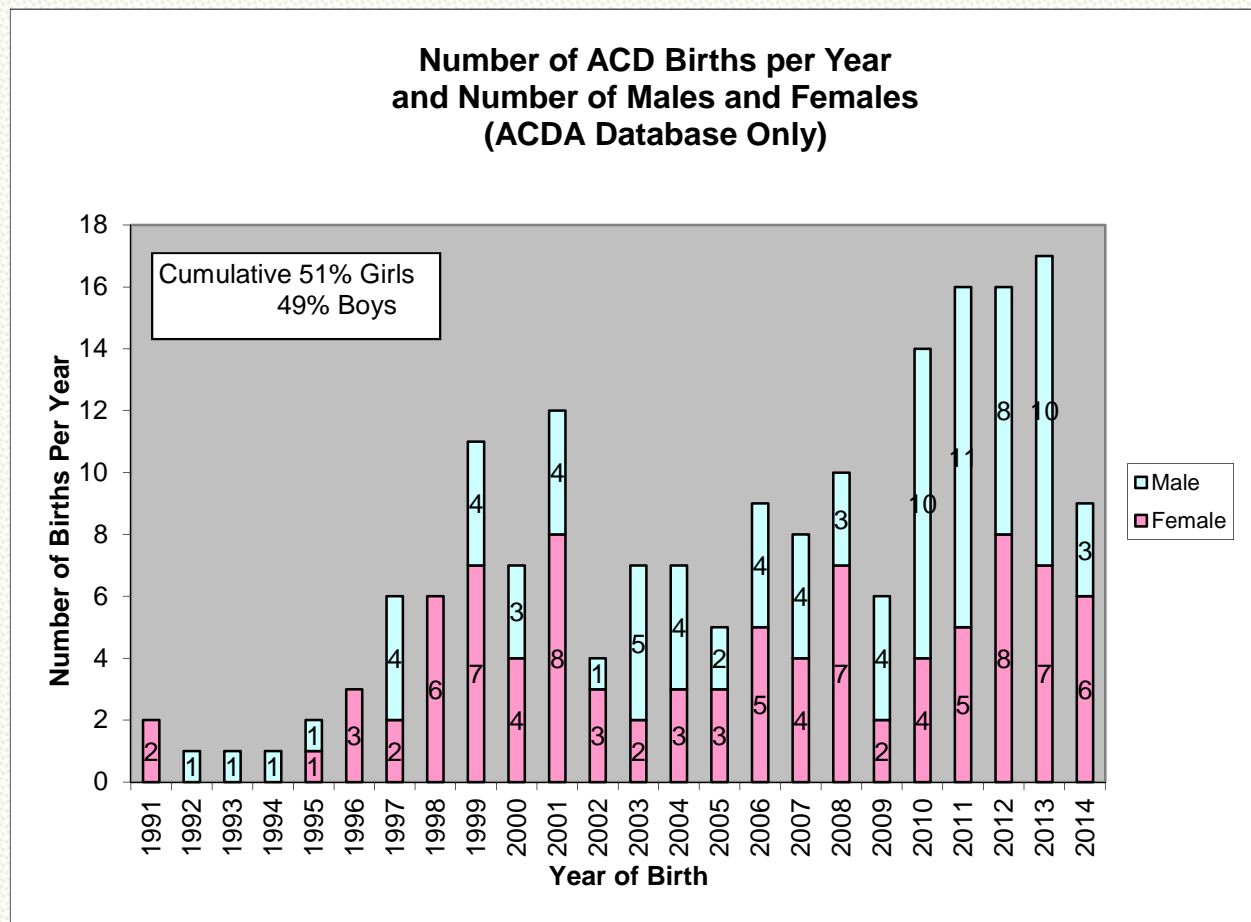
2. What are the chances that our next child will have ACD? This is the most common question we receive and it is a difficult question to answer as all of the cause(s) of ACD are not known. This data is based on the 178 families in the ACDA database:

Families with one child born with ACD	169
Families* with two children born with ACD	3
Families* with three children born with ACD	2
Families with unborn ACD child	4
Families that had a healthy child after an ACD baby **	84
Families who have not had another child (or not known to us)	94

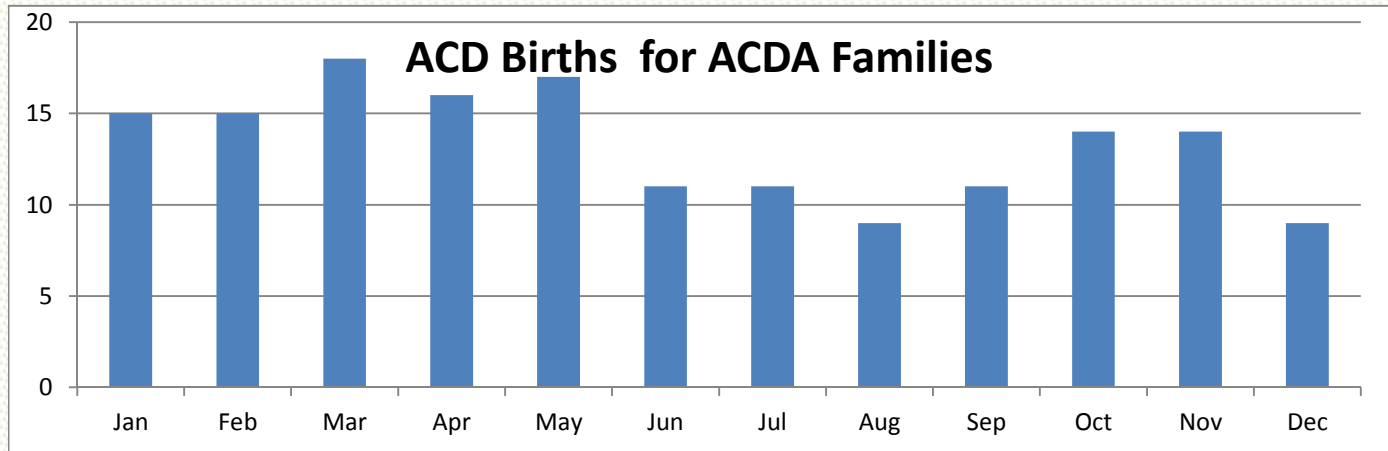
* A Family is defined as children from the same parents (i.e. re-marrying would be another family).

** Two of the four families that had two ACD babies went on to have a healthy child. The other two families with two ACD babies have not had another child together.

3. Does ACD affect males and females equally? The research papers we have seen indicate that ACD 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. Current data shows 51% female and 49% male.

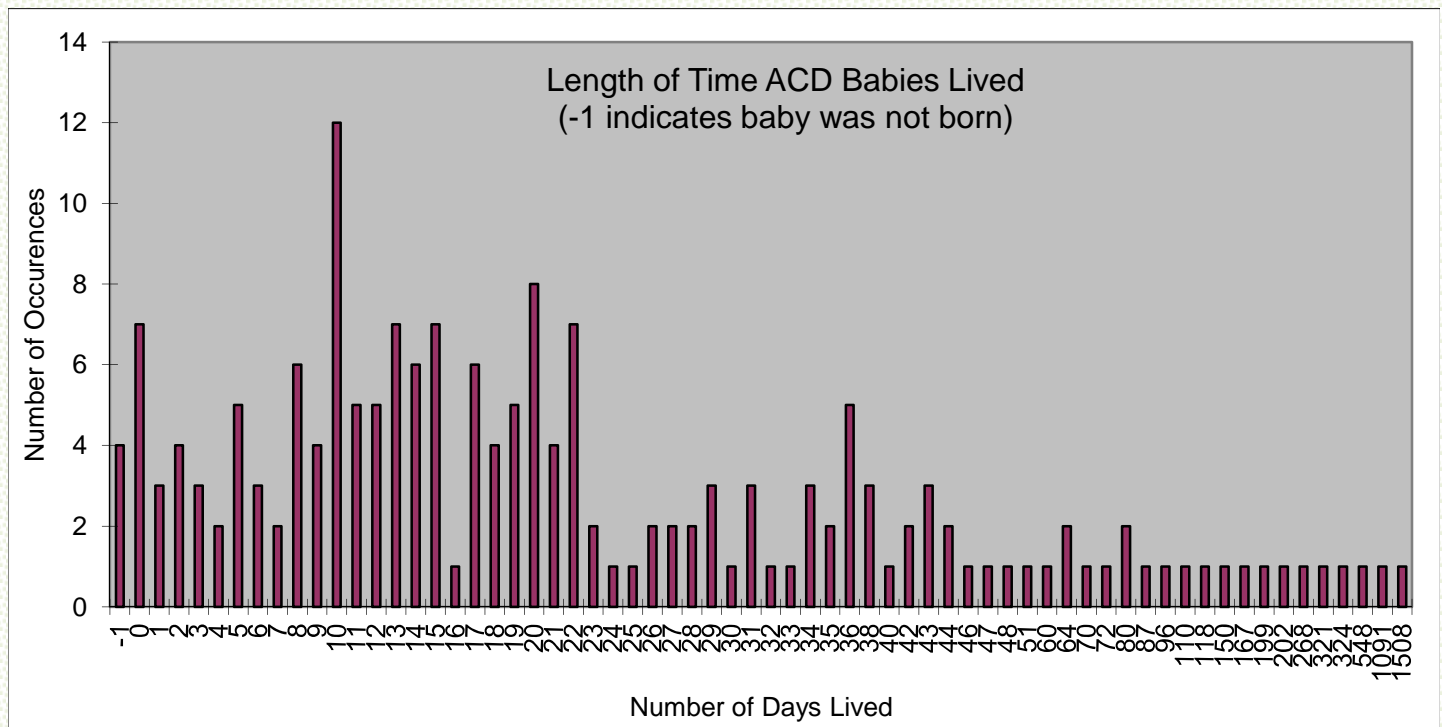


4. Are ACD babies more likely to be born at certain times of the year? Looking at the ACDA family data, there appears to be a minor trend of more babies born in the Spring, but maybe more children in general are born then. The trend is less pronounced now than in prior charts.

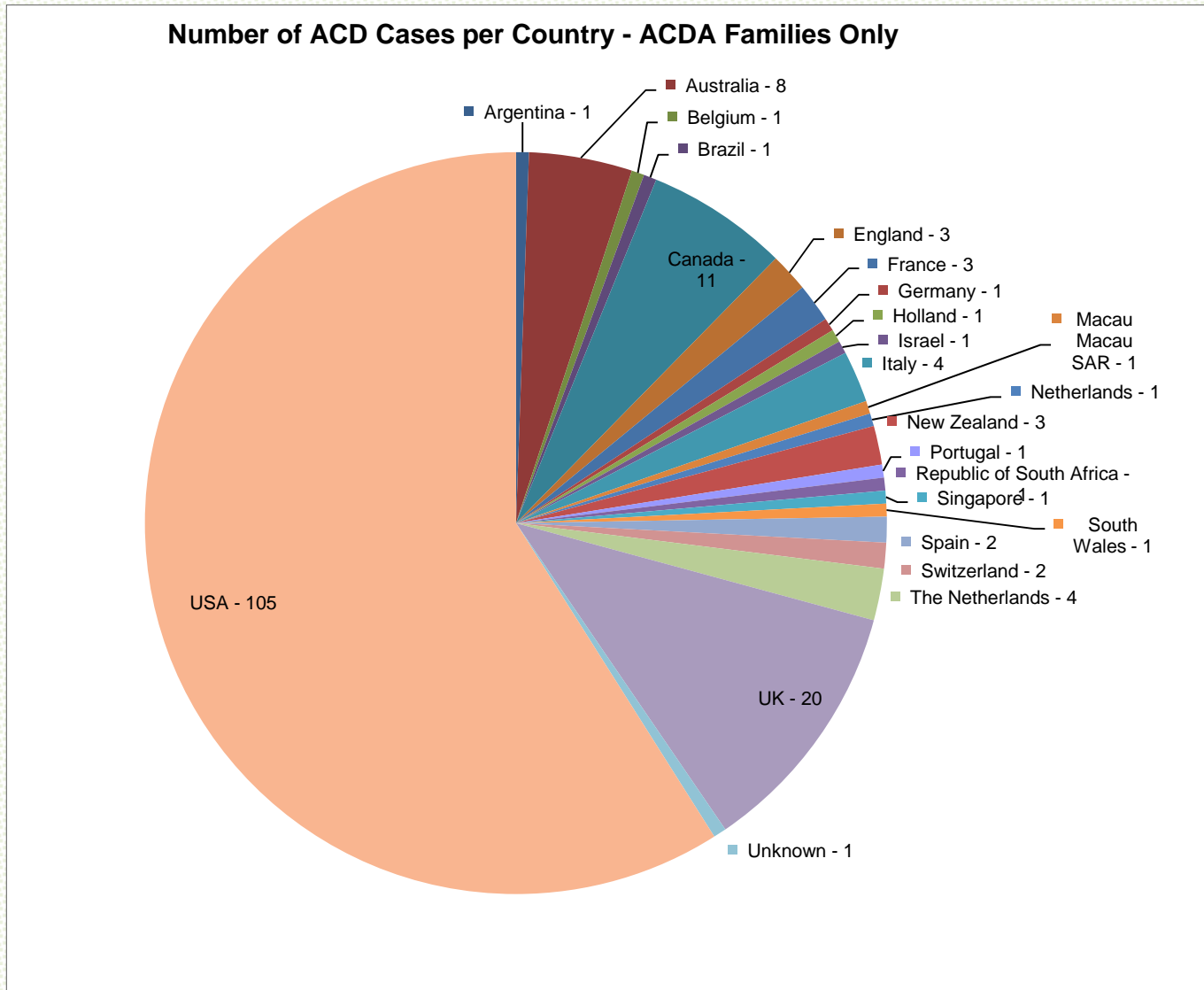


5. How long do babies with ACD live? ACD is a fatal disorder unless heart/lung transplant surgery is performed. Transplants are limited by the short time frame available, limited pool of donor organs and need for matching.

ACD 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 later. When the symptoms present and the level of care available to an ACD baby affect how long the baby will live. We don't have specific data on when babies presented with ACD or what levels of care were provided, but the following chart shows that babies live from less than a day to more than a year in several cases.

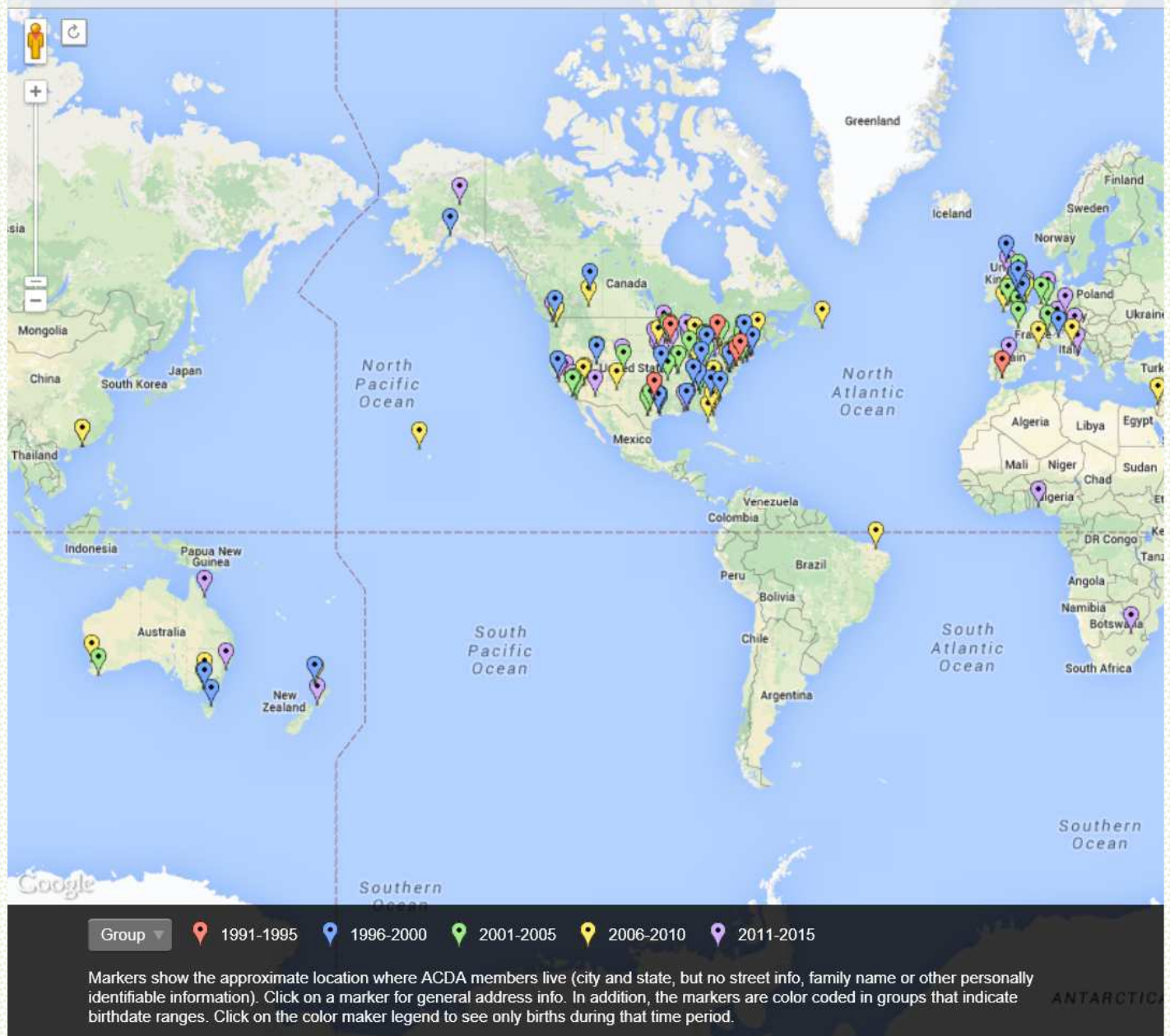


6. Where do ACDA families live? ACDA families are currently located in 24 countries throughout the world, which is an increase of 8 countries since 2012. The largest groups remain the same since the last chart with approximately 59% in the USA, 11% in the UK and 6% in Canada



7. Are there any ACD affected families that live near me? One of the benefits of registering with the ACDA is being able to connect with others that have also experienced the loss of a child to ACD. When you feel all alone and like no one really knows what you've been through, communicating with other members can be very healing. The map below shows ACD family locations around the world. See if anyone lives near you to allow more interaction for support or collaboration on a fundraiser. Consult the ACDA directory for more specific contact information. We can email you an updated directory if you need one.

ACDA Family Locations and Birthdates of Babies



Clicking on the link below will take you to an interactive version of the map above that shows ACD family locations. This will allow you to zoom in and see if anyone lives near you to allow more interaction for support or collaboration on a fundraiser. Consult the ACDA directory for more specific contact information.

<https://batchgeo.com/map/2eccfb200d599775e49bac90325fdae7>

Note: this is an "unlisted" map and only the link above allows access.

Donations Received By the ACDA

Thank you to the following families and friends that have made donations to the ACDA since the Winter 2015
ACDA Notes:

[illegible]

*We are sorry that we do not know the baby that the donation was made in memory of. Please contact us to let us know.

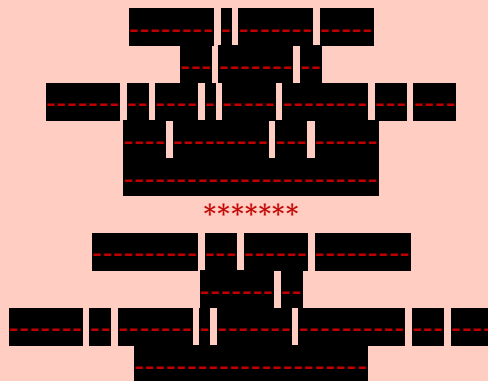


ANNOUNCEMENTS

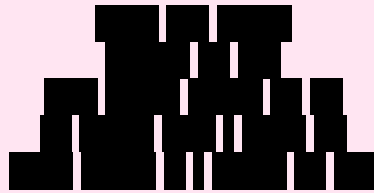
- Amazon Smiles donated \$68.53 to the ACDA based on purchases made during the fourth quarter of 2014 and \$53.90 for the first quarter of 2015. Thanks to all the families and friends that remembered the ACDA when shopping on Amazon Smiles.
- NORD is scheduled to release a Request for Proposal for ACD research by late June 2015. A grant will be awarded later this year for approximately \$55,000.
- Did you know that in 2014, NORD awarded six research grants for four rare diseases totaling \$417,000? Two of the six grants were for ACD research and totaled \$187,000. As such, of the almost 7,000 rare diseases under NORD's umbrella, ACD research grants comprised 45% of the total grants awarded by NORD in 2014.
- The balance of our NORD restricted research fund as of April 30 is \$56,500.
- As of the end of May 2015, the ACDA had approximately \$2,000 in its general fund. Expenses are very few and donations come in on a regular basis. Thanks to all that have donated!

Welcome

Please reach out to our new families and share your story:



Safe Arrivals



Congratulations!



For those of you on Facebook, we encourage you to “like” the ACDA’s public page, which promotes ACD awareness and encourages families to contact the ACDA, (<https://www.facebook.com/ACD.Association>) and to encourage your friends and family to do the same. We’re also happy to “like” the NICUs where your babies stayed in order to help promote the word about ACD.

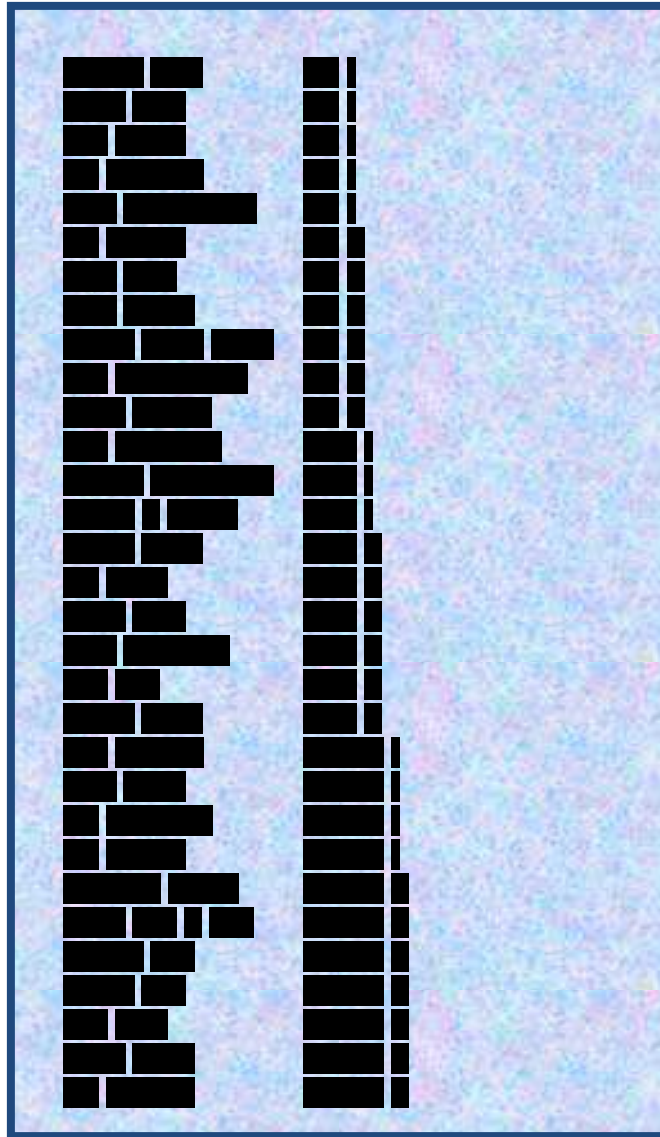
We also encourage ACD parents to join the “ACD Parent Group” (<https://www.facebook.com/ACD.Association#!/groups/168480916544514/>). The ACD Parent Group is a closed group that provides support, information, and a place for families to share pictures and stories of their babies. Contact Emily Eschweiler for more information at Emily_Eschweiler@comcast.net (or search for Emily John Eschweiler on Facebook).



There is an ACDA twitter account for those of you who are on twitter – it is @ACDAssociation. It is ‘manned’ by Amelia Ashwell Lake. The twitter account also tweets what is posted on the PUBLIC ACDA Facebook page. Please be sure to follow us if you are on twitter. Thank you! Amelia

“A family is a circle of love, not broken by a loss, but made stronger by the memories. We remember...”

Remembering Our Babies....



MAKING DONATIONS TO THE ACDA

Now that the ACDA is a non-profit, your donations are tax deductible. There are several ways for you to make a secure donation:

- If you have a PayPal account, the preferred way (to eliminate all transaction fees for the ACDA) is to use PayPal through ebay Giving Works. Go to:
<http://givingworks.ebay.com/charity-auctions/charity/alveolar-capillary-dysplasia-association/69889/?favorite=siteadd&favnpid=69889> Once on the ACDA Giving Works page, select your donation amount on the right side and click DONATE NOW.
- If you don't have a PayPal account, you can use a Credit Card to make a donation. Just follow the 'DONATE' link on our "Donations" page - <http://www.acd-association.org/donations.php> Then, on the lower left section on the PayPal donation page that follows, click the 'Continue' link under **"Don't have a PayPal account?"**.
- Check to see if your company has a Matching Gifts Program. If so, they may match any donation that you make to the ACDA.
- If you prefer, tax deductible contributions can also be mailed to:

The Alveolar Capillary Dysplasia Association
c/o Donna Hanson, Treasurer
5902 Marcie Court
Garland, TX 75044-4958
United States

Checks should be made payable to: The Alveolar Capillary Dysplasia Association.

We will continue to support and contribute to our ACD Restricted Research Fund at NORD. Until the ACDA can assemble its own Medical Advisory Board, we will have NORD continue to issue research RFPs and grants on behalf of the ACDA.

Please remember that when you make a donation to NORD, earmark the donation to our restricted research fund by writing on your check (or enclosed note) that your donation is for our account. Otherwise, the funds will go to NORD's general fund. See page 11 for how to make a donation to NORD.

Make a Tax-deductible Contribution to NORD for ACD Research

In the spring of 2002, the ACDA established an ACD Research Account at NORD. This means that your contribution to NORD can be earmarked specifically for ACD research. As stated below in *NORD's Rare Disease Clinical Research Program Policy*, NORD requires that a research account reach \$33,500 before it will initiate the grant process to award research money to the medical community. Therefore, the goal of the ACDA is to raise more than \$33,500 for research.

To make a tax-deductible contribution to NORD for ACD research either by mail or on the NORD website, please use one of the instructions:

*** Make a Donation by Mailing a Check ***

- Please make your check payable to "NORD - Alveolar Capillary Dysplasia Restricted Research Fund" to earmark your donation for ACD research.
- In the memo section of the check or on a separate note attached to the check, state that the donation is "in memory of (name of child)."
- Your family and friends can attach a note to their check with your name and address and NORD with promptly notify you of their gift.
- Send your check to the following address:

National Organization for Rare Disorders, Inc.
P.O. Box 1968
Danbury, CT 06813-1968 USA

The most critical part of this process is ensuring that your check is made out to "NORD - Alveolar Capillary Dysplasia Restricted Research Fund" to ensure that your donation is earmarked for our ACD Research Account.

*** Make a Donation on the NORD Website ***

Go to <https://www.rarediseases.org/about/support/research-donations>. Select "Alveolar Capillary Dysplasia" in the research fund pull-down menu and complete the rest of the form. In the "Additional Comments" box, type "Alveolar Capillary Dysplasia Restricted Research Account."

Special Information for Families Living Outside of the United States

NORD recommends that families living outside of the United States use a credit card to make a donation since it costs less to convert international currency when using a credit card. Use the NORD website at <https://www.rarediseases.org/about/support/research-donations>.

Contact Information

Steve Hanson, President - president@acd-association.org
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Donna Hanson, Treasurer - treasurer@acd-association.org

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