

ACDA NOTES

From The Alveolar Capillary Dysplasia Association

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

Summer 2013

Dear Friends and Family,

We have many new members that have registered with the ACDA since the last newsletter. Please take a few minutes and contact them. We know that most of you have found some level of comfort knowing that there are other families that experienced the same depth of loss as you. Our new families experience that same feeling.

Our European families are making us in the US look bad when it comes to fundraising! If we are going to make a difference in research, we all need to be actively fundraising. It's a challenge but we hope you find a way to make a difference.

Fondly,

Steve and Donna Hanson
Executive Directors ACDA



Raising Awareness



Diana Locke, mom to Christopher, continues in her ACD awareness campaign for medical students at the University of Florida Gainesville. With an official syllabus for the Genetics and Health class that includes a case presentation on ACD, Diana lectured to 130 medical students on August 28. The students' final exam was given after her presentation for the first time so ACD was included in that final exam for the Genetics class. Looks like this will be an annual presentation that Diana will make to ensure that future doctors are aware of ACD.

Great work Diana!

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Update on NORD Grants

NORD has received Dr. Szafranski's interim progress report for his study entitled, *Long Non-Coding RNAs as Potential Diagnostic and Therapeutic Targets in Patients with Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins (ACDMPV)*. Dr. Szafranski received outstanding reviews from NORD's Medical Advisory Committee for the entire, in-depth interim report. Dr. Szafranski's report is below but is preceded by a synopsis in layman's terms provided by Dr. Simon Ashwell, dad to David. Thanks Simon!!!

Synopsis of Dr. Szafranski's Report

Abnormalities in the *FOXF1* gene are known to result in ACD. The goal of Dr. Szafranski's research project is to learn more about how the *FOXF1* gene is regulated (controlled). Knowing more about how *FOXF1* works should allow him to search for other genetic mutations that can cause ACD.

Dr. Szafranski has so far identified several pieces of genetic code (called lncRNA's) that are located close to *FOXF1*, are expressed in lung tissue, and which might therefore regulate *FOXF1* expression. He is now conducting experiments to see which of these lncRNA's regulate *FOXF1* expression most clearly. He then plans to study the mechanisms by which lncRNA's regulate *FOXF1*.

He hopes that this work will increase the number of identified genetic abnormalities that have been found to cause ACD and thus assist in prenatal diagnosis and perhaps even treatment for ACD.

Original Report from Dr. Szafranski

Loss-of-function mutations or deletions of *FOXF1* can result in development of ACDMPV. Knowing how the expression of *FOXF1* is regulated would not only define new targets for more effective prenatal screening for pathogenic mutations and CNVs, but would also facilitate development of therapies for ACDMPV cases that result from *FOXF1* haploinsufficiency.

During the first few months of our research on ACDMPV sponsored by NORD, I identified several non-protein coding transcripts called long non-coding RNAs (lncRNAs) that, based on the proximity of their genes to *FOXF1* and their lung-specific expression, might regulate *FOXF1* expression in human fetal lungs. Recently, lncRNAs emerged as key regulators of gene expression that function as scaffolds for chromatin-modifying complexes or as direct transcriptional modifiers. It is worth mentioning that only 1.5% of the genome is responsible for protein coding. However, the majority of the remaining portion of the genome is still transcribed producing RNAs that are not translated to proteins, but as, e.g. lncRNAs, may play critical role in regulation of the expression of protein-coding genes.

I am currently conducting the experiments that will show which of the identified lncRNAs increase or decrease the expression of *FOXF1*, and which of them function most effectively. Knowing this, I will then elucidate the molecular mechanisms of this regulation, starting from identification of protein complexes interacting with these lncRNAs. This part of the research is anticipated to significantly broaden the list of chromosomal loci for diagnostic screening for ACDMPV-causing mutations and CNVs and will likely define new targets and strategies for effective ACDMPV therapies.

In Memory Of...

With grieving, heavy hearts we are saddened to share with you the passing of two precious babies of ACDA members. We know that we speak for all the members of the ACDA when we express our sincere condolences and sympathy to these two families.



■■■■■, daughter of ■■■■■, was 18 months old when she passed away at St. Louis Children's Hospital. As a result of having ACD, she had received a double lung transplant in April but passed away from respiratory distress. You can read more about Omari at:



■■■■■, son of ■■■■■, was 10 months old when he passed away in Chattanooga, Tennessee from complications unrelated to ACD. Below is some information about ■■■■■ taken from the Chattanooga Funeral Home and Chapel.

■■■■■, 10 months, of Chattanooga passed away Saturday, ■■■■■. He had the biggest and brightest smile of all time, everything made him giggle. We were able to catch him on video when he surprised us by saying mama. He would jibber jabber from crib to crib with his brother ■■■■■. He loved to pull up and walk holding on to the furniture or someone's hands. ■■■■■ loved to eat cheerios especially when his brother was feeding him. Peek-a-boo and tickles were his some of his favorite games, as well as playing cars with his brother and watching Barney. For such a short life He knew how to bring joy to everyone he was in contact with. ■■■■■ loved with all his might. He totally looked up to his brother, ■■■■■ and sister ■■■■■. He was preceded in death by a brother ■■■■■ and a sister ■■■■■. Survivors include his parents ■■■■■; brother ■■■■■; sister ■■■■■; grandparents ■■■■■ and ■■■■■; aunt ■■■■■; the ■■■■■ family, Godmother ■■■■■ and Godfather ■■■■■, family pet Shaddow and many more relatives and friends.



Trigger Music



In the last *ACDA Notes*, a number of parents on the ACD Parent Group on Facebook shared the songs that remind them of their child; songs that they listened to while pregnant, played at their baby's bedside or as part of their memorial service. Below are some additional songs that have special meaning to other ACDA families:

Someone Like You by Adele

We have so many songs. The main one, which might sound strange is Adele's "Someone Like You". It was very popular at that time and I specially remember it coming on the radio (we had one in her room) numerous times and I would sing whatever was on to her. Many Coldplay songs too, especially "Paradise" which reminds us of her running around happy in Heaven. Also a sweet song called "Unknown Brother" by The Black Keys. It's about the singer's brother-in-law who he never met because he died from cancer as a teen. It is a really cool song and made me think of hopefully getting to share it with any siblings we might be blessed with. One of our biggest is Jesus Loves Me, we sang it to her as she left us and at her funeral.

Why Does My Heart Feel So Bad by Moby

We had a little bear sitting next to Jasmijn while she was in the hospital that played "Somewhere Over The Rainbow" from the Wizard of Oz. We played that at her funeral. But the song that gives me tears the most is "why does my heart feel so bad" by moby. I listen to all the songs a lot. Although it makes me cry, it feels good at the same time, remembering her.

Hello, Goodbye by Michael W. Smith

We had 'Noah hello, goodbye' by Michael W. Smith at our son's service, at the time I listened to it non-stop but now I can't listen to it at all. The lyrics are also part of the wording on his headstone.

Return to Pooh Corner by Kenny Loggins

Blessings by Laura Story

It ministered to my heart throughout my pregnancy, but it wasn't until her birth that I would understand just why.

Bend and Break by Keane

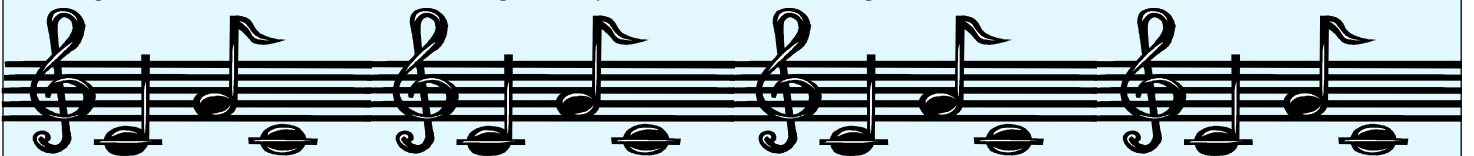
We played Keane's album, *Hope & Fears*, when our son was being born. A number of tracks from that bring back memories of his time in this world but "Bend and Break" is one of the main ones.

Daylight by Maroon 5

"Here I am staring at your perfection, In my arms, so beautiful. The sky is getting bright, the stars are burning out, Somebody slow it down. This is way too hard, cause I know, When the sun comes up, I will leave. This is my last glance that will soon be memory." We also listen to "If I Die Young," by The Band Perry. "Lord make me a rainbow, I'll shine down on my mother, she'll know I'm safe with you when she stands under my colors."

Miracle by Celine Dion

Celine Dion's song "Miracle" was THE song. I played it at home for my daughter, all the time. Also played it through the whole wake. If I need a good cry, I listen to that song.



Fundraising NEWS

By Emily & Tim Eschweiler
ACDA Fundraising
Committee Chairpersons

Update from The David Ashwell Foundation

Amelia Ashwell Lake and Simon Ashwell
Parents of David
June 2013 – September 2013

Donations to ACD Research via The David Ashwell Foundation have been continuing. Since March 2011, we have raised £119,411 (\$187,833) for ACD Research. £33,118.00 (\$52,053) was transferred in February to NORD and over £86,293 (\$135,738) has been raised **since** the NORD transfer in Feb 2012. To have raised over £119,000 in memory of David and other babies is beyond what I could have imagined.

The fundraising by friends and families of children lost to ACD is the only way money will be raised for research into this disease we all know too well. This amazing amount is the result of a great team effort from families across the UK and Europe. This year a lot of money has been raised in memory of other ACD babies and not just David.

- In August, the second charity car show was held in our small town of Stokesley. It was in aid of The David Ashwell Foundation and Tommy's (Baby Charity). The event was well attended and raised a fabulous £2575 (\$4,050) for our charity. See photos on the next page of cakes baked by my friends to sell at our cake stall and the event flyer/poster.
- As you know, Jo Taylor (mum of Alexander) nominated The David Ashwell Foundation as Thomson Airline's local charity. This donation route has now raised over £32,000 (\$50,336).
- Friends of Ellie Merritt and Nathan Flores (parents of Noah) organized an event for The David Ashwell Foundation.
- Stamp donations continue to come through! Thank you all for your contributions.

All UK and 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](http://DavidAshwellFoundation)
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](http://DavidAshwellFoundation)

For additional information, please contact Simon and Amelia [http://David Ashwell Foundation](http://DavidAshwellFoundation)
davidashwellfoundation@yahoo.co.uk ; 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. All money raised will be transferred to NORD.

*Photos from
The David Ashwell Foundation
Fundraiser
Charity Motor Show
August 2013*



Cakes baked for the fundraiser



Guest Host Steve Furnell
from TFM's The Steve & Karen Breakfast Show!

AUTO BODYCRAFT
STOKESLEY

IS PROUD TO PRESENT THE 2ND ANNUAL

CHARITY MOTOR SHOW

SUNDAY 11th AUGUST 2013 10.00am - 3.00pm

Admission: £2.50 Over 60's & 12-18 year olds £2.00 under 12s FREE

Venue: Terry Dicken Industrial Estate, Station Road, Stokesley.

Huge range of exciting and interesting vehicles on show, plus much much more!

- Classic & Vintage
- Supercars
- Sport, Modern
- 4x4s
- Bikes & Scooters
- Mountain Bike Skills
- Unusual & quirky
- Stalls • BBQ
- Raffle
- Live Music
- Ample Free Parking
- Obstacle Course.

.....and more!

Raising funds for...

The David Ashwell FOUNDATION
Registered charity No. 1121783

Tommy's
Funding research into stillbirth, premature birth and miscarriage, and providing information for parents-to-be.
Registered charity No. 1060508

The 2012 Charity Motor Show raised £5027 for the David Ashwell Foundation.
Sincere thanks to everyone who contributed.

If you have a motor of interest that you would like to include in our show, please call Alex
01642 715894 or 07738 430750

Supported By:

ZENITH LEISURE
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www.DARRENALICKCARBS.co.uk

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www.autobodycraftstokesley.co.uk

A Note From Emily - In 2010, when Donna and Steve asked if I would take on the project of car magnets, my initial inclination was to say no. I was still dealing with my grief of losing Joey, and Ava was on the way. I couldn't imagine how I would find time to research the project, order the magnets, collect orders from people all over the country, and get the magnets shipped. However, as I got into the project, finalizing Raquel's design, working with a graphic artist to make sure that the image would work for our project, and finding a company where I could buy not only car magnets, but fridge magnets as well, something magical happened. I started to feel like I was making a difference. Not only would the magnets raise awareness globally, but the proceeds would fund much needed research. We have not had many fundraisers in the U.S. I know that as busy, grieving parents that sometimes the thought of one more item on our plate is hard to imagine. However, the rewards can be great too.

Even if you have never held a fundraiser before, I encourage you to consider it. The outpouring of support for the ACDA community to the magnet project was overwhelming and healing.

Thomson Airlines Fundraiser

By Jo Taylor, Mom to Alexander

West Sussex, England

As you all know, my work Thomson Airways collects for the David Ashwell Foundation on every flight back into London's Gatwick Airport. We mainly do short haul flights to Europe but also have flights to Sanford, USA, Cancun, several Caribbean islands, Kenya and the Maldives. We have also become the first UK airline to fly the Dreamliner, so it is a pretty big company in England.

My colleagues voted in our charity in March 2012 and the first collections started that May. The Crews have been collecting ever since and although the collections were supposed to be for a year, ours was extended until October 2013 to bring it in line with the Financial year (so an extra 4 months of money).

Throughout this time, the summer months carry the most amount of passengers and this has been reflected in the amounts raised. We receive a quarter of all of the monies raised with the other quarter going to Born Free Animal charity and the rest going to the Family Association who look after disadvantaged children.

In the first year (May to May) just under £25000 was raised and up until July 2013, over £32000 has been donated by our passengers. August's total will be the last one as in September all monies collected are going to Breast Cancer. So I will update you all on that amount, once it's in.

All in all a huge achievement from all my friends and colleagues who read the charity talk and collect the monies which has been a lovely tribute to Alex and all our ACD Angels. Gatwick has historically not had the most generous of passengers for charity donations, but they gave and proved us wrong over the last 15 months!!

The monies raised will go towards years of future research which will hopefully lead us nearer to the answers we are all after.



Fundraising Events from Ellie & Nathan Flores

Parents of Noah

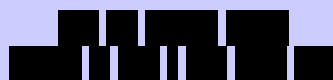
Surrey, England

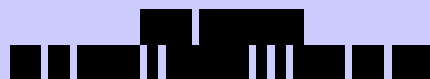
Family friends of ours help to run our local steam show every year that is held over the first weekend in August. The charity the steam rally raises money for has been Canine Partners for many years and our friends fought very hard to try and get this changed to The David Ashwell Foundation. In the end it was decided that the funds raised would be split between the two charities. We are unsure at the moment as to how much had been raised. Also another family friend has continued to raise money at car shows over the summer with her bucket and 'Nuggets for Noah'.

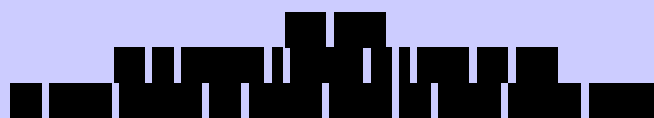


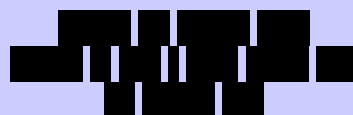
Please Welcome Our New ACDA Families:









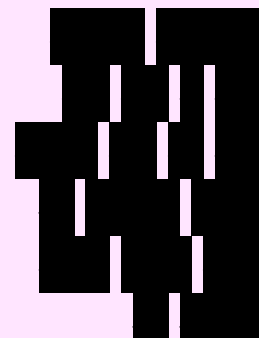
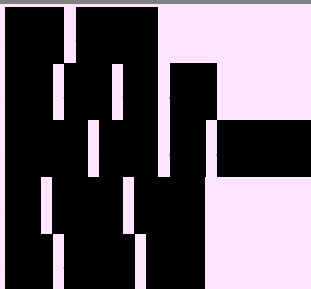




- Thanks to Verónica Liaudat of Argentina and mom to Uma, who found a link to a report on ACD published in January 2013. This is a case study from Switzerland and while it doesn't present any new findings on ACD, it is continuing evidence that ACD is becoming better known in the medical community throughout the world.
<http://www.neonet.ch/assets/pdf/2013 - Interesting cases - ACD.pdf>
- Thanks to Verónica as well as Karin Titze-Frech of Germany and mom to Jannick for their efforts to find new information and articles about ACD on the web!



Safe Arrivals



Make a Tax-deductible Contribution 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>.

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