ACDA NOTES

From The Alveolar Capillary Dysplasia Association

http://www.acd-association.com

Winter 2015

Dear Families and Friends,

Because of generous donations from families and friends as well as very successful fundraisers, we hope to have another grant for ACD research awarded by NORD in 2015. The deadline for making a donation to NORD is March 1. Even a small group of people can effect change. Many thanks to all of you that have contributed or held fundraisers!

February 28 is Rare Disease Day, an international day to recognize rare diseases and their impact on global health. There are many ways for you to participate and honor your baby. I hope that you will join the ACDA in raising awareness of ACD and other rare diseases on February 28. Check out the information on Rare Disease Day on page 8.

As you will read in this newsletter, we have many families doing amazing things to raise awareness and funding for ACD research-all in memory of their baby. If you too are making a difference, please share your stories with us so that your efforts can inspire someone else. The ACDA would love to hear from you.

Fondly,

Steve Hanson ACDA President

Father to Eric – June 7-17, 1997

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Employer Matching for Tax Deductible Donations

By John Rista, Fundraising Committee and father to Johnny

If your company has a matching gift program in the United States, now is a great time to take advantage of such a program since the ACDA was granted 501(c)(3) status by the IRS!

As an employee of Bank of America (BOA), I was pleased to discover there are two ways in which my donations to the ACDA are matched by my employer. The first is where they match my monetary donations dollar for dollar up to \$5,000 per calendar year. It was a very quick process to obtain the dollar for dollar match. I simply went to our internal philanthropy website, searched for the ACDA in Texas, answered a few simple questions and gave the amount and date of my donation. My employer then gave me a receipt that I submitted to the ACDA via email to treasurer@acd-association.org. The ACDA then confirmed the donation and they will receive the matching gift from BOA.

The second way my employer provides matching gifts is via a volunteer grant where I record my time volunteered to the ACDA. If I volunteer 50 hours in a year, my employer will give a volunteer gift of \$250. If I volunteer 100 hours or more, the ACDA will receive a volunteer grant of \$500. Fortunately, Bank of America provides flexibility with the option of matching monetary contributions and/or time.

If you are uncertain if your company has any type of matching gift program, reach out to your employer's Human Resource department to find out any details and requirements. If your employer has a matching gifts program for charitable organizations, your contributions to ACD research can easily grow!







Humana also has a similar matching gift program as Diana Locke, mom to Christopher, recently discovered. Through the Humana Foundation's Giving Together matching gift program, the Humana Foundation matches associate donations to approved 501(c)(3) nonprofit organizations up to \$100 per calendar.

Because of John and Diana's actions, the ACDA recently received donations from Bank of America and Humana!

Research Updates

Summary of Recent ACD Research from Baylor College of Medicine

"Molecular and clinical analyses of 16q24.1 duplications involving *FOXF1* identify an evolutionarily unstable large minisatellite"

Avinash V. Dharmadhikari I, Tomasz Gambin, Przemyslaw Szafranski, Wenjian Cao, Frank J. Probst, Weihong Jin, Ping Fang, Krzysztof Gogolewski, Anna Gambin, Jaya K. George-Abraham, Sailaja Golla, Francoise Boidein, Benedicte Duban-Bedu, Bruno Delobel, Joris Andrieux, Kerstin Becker, Elke Holinski-Feder, Sau Wai Cheung, Pawel Stankiewicz

Summarized by Dr. Simon Ashwell, father of David

Mutations or deletions of the FOXF1 gene cause ACD. The effects of, in effect the opposite, duplications of FOXF1 are unknown. This study investigated the outcomes of FOXF1 duplication. Four unrelated people with duplication of FOXF1 were identified. The first, a 4 ½ year old boy has speech delay, behavioral issues and facial changes but he also has a further genetic abnormality that may account for these. The second, a 13 year old boy, has autism, behavioral problems and limited growth. He has two other genetic abnormalities that again may account for some of these issues. The third patient, an adult, has pyloric stenosis and other gut abnormalities. She has a daughter with similar gut abnormalities to whom she has passed the FOXF1 duplication. The fourth is a 10 ½ year old boy with speech and motor delay and mild learning difficulties.

None of the four have any lung abnormalities. All four have duplications of FOXF1. The first three patients inherited the duplication from healthy fathers. This is consistent with the previous evidence that FOXF1 is paternally imprinted, i.e. the gene inherited from the father is inactive; only the gene inherited from a mother is expressed. However, the third patient inherited the FOXF1 duplication from a healthy father suggesting that FOXF1 is paternally imprinted in the lungs only, as she had gut abnormalities. In the fourth patient, the duplication arose *de novo* on the maternal chromosome.

The study shows that duplication of FOXF1 does not cause any lung abnormalities but can cause gut abnormalities.

Congratulations NORD Awards Two Grants for ACD Research

In January 2015, the ACDA and The David Ashwell Foundation were notified by the National Organization of Rare Disorders that they had awarded two grants for ACD research. The funding for these grants was raised and donated by family and friends of the ACDA and The David Ashwell Foundation.

One of the grants will support Dr. Csaba Galambos, a pediatric pathologist at Children's Hospital Colorado and the University of Colorado Denver. The project, "Role of Pericytes and Platelet-Derived Growth Factor-Beta Signaling in the Pathogenesis of Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins," is an integrative approach utilizing banked human ACDMPV lung tissues, in vivo animal modeling and novel in vitro cell-based methods, to determine whether upregulation of PDFG-B signaling is sufficient to cause defective pulmonary vascular remodeling and PHT, and to identify PDGF-B-driven mechanisms that are responsible for impaired angiogenesis and defective lung growth in ACDMPV. Dr. Galambos studied under Dr. Claire Langston, the foremost pathologist on ACD who has since retired.

The other grant will support Przemyslaw Szafranski, Ph.D. Assistant Professor of Molecular and Human Genetics at the of Baylor College of Medicine. His project, "Towards designing ACDMPV therapy: Deciphering epigenetic regulation of FOXF1," will identify key IncRNAs and their interactors involved in epigenetic regulation of FOXF1, decipher the mechanism of how the expression of FOXF1-controlling IncRNAs, in particular those involved in FOXF1 silencing on the paternal chromosome, is regulated, and identify additional FOXF1 genes involved in the etiology of ACDMPV.

Congratulations to these two researchers! We are grateful for their continued support of ACD research.



Report from The David Ashwell Foundation

From Amelia Ashwell Lake, mother to David

Our balance on 31 December 2014 was £7997 (\$12,064). This money will be transferred to NORD for the next ACD research grant (before March 2015).

Since March 2011, we have raised £136,127 (\$205,368) for ACD Research. The David Ashwell Foundation has contributed to two recent grants for ACD research issued by NORD; £33,118 (\$52,053) was transferred in February 2012 and over £95,011 (\$157,072) was transferred to the NORD ACD account in March 2014.

This amount has been raised by the ACD families in the UK and Europe plus their families and friends. A recent example is the fundraising by the Davey family who has recently lost their beautiful daughter Hope to ACD (please see Kerry's update). Also thanks to the Kinchington Family in Scotland for a generous donation from a raffle they had for The David Ashwell Foundation in November.

Fundraising events that have occurred recently include an Usborne book party I held in my home in November which raised just over £300 (\$452). My sister, Anisa, had a stall at the Dubai Flea Market in December, selling clothes her friends had donated. She raised over £500.00 (\$754). My mum continues to raise money by crocheting baby blankets. Over Christmas we had numerous generous donations in lieu of Christmas cards (over £500).

Future fundraising includes Simon, his sister and some friends running the Edinburgh Marathon for The David Ashwell Foundation in May 2015. If anyone in UK would like to support us or join the team, please get in touch.



Amelia's sister, Anisa, and friends raising money at flea Market in Dubai

UK families - please don't forget to use The Giving Machine when you shop!
Using this has raised £694 (\$1047) since 2011 and in Germany the equivalent website (thanks to Karin!). In December, we were even featured in The Giving Machine's media campaign as we have raised so much money with them! 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 familes 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

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 for ACD Research.

Fundraising News

Go Fund Me Fundraisers

Two families have been busy raising money for ACD research in memory of their babies. Their efforts are truly inspirational acts during a time of overwhelming grief of having lost a baby. Be sure to visit their Go Fund Me site to read their stories, view some emotional pictures and be inspired.

In Memory of Dylan Jenks:

\$25,360 has been raised by the family and friends of Dylan Jenks. Parents Jason and Sophia share their story of their son, Dylan, at http://www.gofundme.com/thedylanjenksfoundation. Their mission is "to make significant ongoing donations in order to fund ACD research in loving memory of our beautiful son, Dylan Aaron Jenks. We also want to make a substantial donation to the Cardiac Intensive Care Unit (CICU) at Children's National Hospital in Washington DC in order to help fund the Beads of Courage program, as well as other amazing programs available to other parents like us at the CICU."

In Memory of Sadie Rose McCasland:

\$3,960 has been raised by Sadie Rose's family and friends. Parents Candice and Bryan McCasland have amazing photos and a beautiful story about their short time with their daughter, Sadie Rose. Their goal was to raise \$3,000 to support the 2015 grant at NORD – which they have exceeded! http://www.gofundme.com/SadieRoseMcCasland

ebay Post Week - First Week of March

Do you have household items sitting around the house you don't use anymore? Have your kids outgrown some of their toys? Ever received a gift that you have never opened? If so, do some spring cleaning and sell those items on ebay. Join the ACDA in selling some items the first week of March to raise money for ACD research. The following steps outline an easy way to designate the ACDA as your charity of choice when you sell on ebay and to choose a percentage of your proceeds to donate to the ACDA.

So, Step 1 - Choose the ACDA as Your Charity

- Add to my charities

 Go to http://givingworks.ebay.com/charity-auctions/charity/alveolar-capillary-dysplasia-association/69889/
- Click on the red heart "Add to my charities"
- You will then be asked to log into your ebay account

Step 2 – When you are ready to sell on eBay:

- To sell an item for Charity you will need to SWITCH TO ADVANCED TOOL
- IMPORTANT NOTE Remember to switch to the advanced tool every time you sell to support charity, so you'll always see the option to support your cause.
- After setting the price of your item, select the ACDA as the non-profit you want to support and choose the percentage amount you would like to donate
- Your item will stand out to buyers with a blue and yellow ribbon logo displayed right next to the item's title. Charity listings on eBay have up to 30% higher sell through rates then non-

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Charity items and they sell for between 2-6% higher prices.

- After the sale ends, sellers pay their donation to PayPal Giving Fund through their eBay Donation Account. You can actively pay the donations or wait four weeks for PPGF to collect the donations automatically.
- Once you sell your charity items, you'll receive listing fee credits for the same percentage amount you donated.
- PayPal Giving Fund processes the donation, distributes tax receipts and pays the donation to the ACDA in the monthly donation payout.

For more information for sellers, go to http://givingworks.ebay.com/charity-auctions/charity-sellers/

Start your spring cleaning while the weather is keeping you inside and prepare to sell! Be sure to share this fundraising idea with your friends and family!



Fundraising Idea

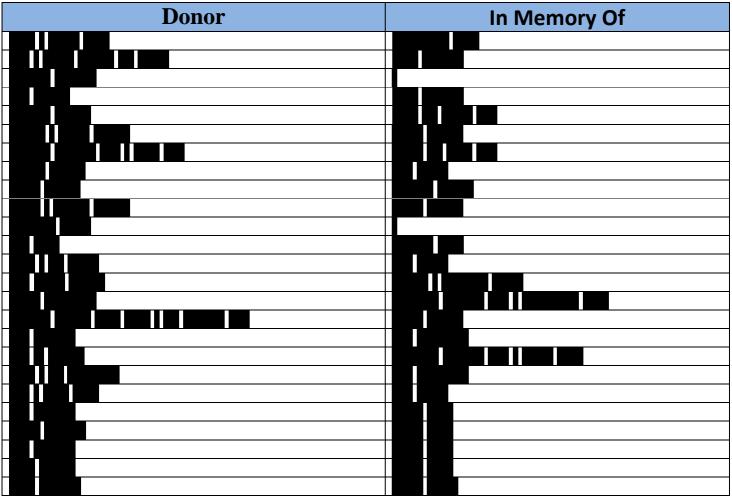
Change for Charity

Start a "Change for Charity" fundraiser in the comfort of your home. Place a jar in your home and empty your spare change in it. Visitors to your home may want to donate their coins also. When you have collected a significant amount of change, take your coins to the bank and make a check out to the ACDA. This is a great way to get your children involved in charitable giving and foster a "giving" heart.



Donations Received By the ACDA

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



^{*}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

Quarterly ACDA Board Meeting Held

The quarterly Board Meeting of the ACD Association was held on December 28, 2014. The following highlights the discussions of the Board:

- Eliza Rista, Emily Eschweiler and Diana Locke were confirmed as members of the Communications Committee. One of the responsibilities of the Communications Committee will be to review and recommend new ACDA logos to the Board.
- John Rista was confirmed as a member of the Fundraising Committee.
- The Treasurer reported that \$2,628 had been received in donations through Paypal, Paypal Giving Fund or checks. In addition, a check from Café Press for \$53 had been received. Donations from Amazon Smiles are expected in late February.



ANNOUNCEMENTS

- The ACDA website has been changed to a .org. Be sure to change any links you have to acd-association.org.
- The NORD balance of the ACD Restricted Research Fund as of January 31, 2015 is \$32,418. The
 deadline is March 1 to make a donation to help us reach our goal of \$33,500 for another grant in
 2015! Thank you!!!
- mom to Hope, is raising money for ACD research by running in a marathon in March.
- Several designs for a new ACDA logo were submitted and are under consideration by the Communications Committee and the Board of Directors. Many thanks to the following people that submitted designs:
- Our CafePress site has products your can purchase (t-shirts, mugs, etc.) with the current ACDA logo and the ribbon! We receive 10% of the purchase price (less fees), so encourage your friends and family to support this fundraiser for the ACDA. Check out our site at http://www.cafepress.com/acdawareness.
- The ACDA Board of Directors has authorized a \$3,000 donation to the ACD Restricted Research Account at NORD. This was made possible by families and friends that made donations to the ACDA since August 2014. Thank you!



Celebrate Rare Disease Day



Rare Disease Day is an international advocacy day to bring widespread recognition to rare diseases as a global health challenge. The day is celebrated on the last day of February every year - February 28 in 2015. Anyone can be involved in Rare Disease Day and there are many suggested activities. The day has been established as a grassroots advocacy day and we encourage everyone to participate in some way! Learn more about the History of Rare Disease Day, Past Successes or the Goals and Plans for this year's campaign. See more at: http://rarediseaseday.us/about/

Some ideas for ways for you to get involved:

- Write your representatives to tell them why rare diseases are important to you
- Become an individual supporter to receive updates about the day
- Participate in the Handprints Across America project Plan or participate in a State House event
- Write a letter to the editor and send it to your local paper about what Rare Disease Day means to you
- Send a proclamation request to your representatives
- Organize an awareness display at a local school or hospital
- Bring Rare Disease Day into a local school Host an event at a local community center or library
- Create artwork to commemorate the day and share it with us on Facebook
- Buy Rare Disease Day merchandise, such as t-shirts and mugs, to help make people aware of the day
- Participate in an event near you
- Share information about Rare Disease Day on your social sites

Check out this link where they have sample letters and more details on the suggestions listed above: http://rarediseaseday.us/take-action-now/suggested-activities/



When we had our first daughter in 2008, we couldn't have been happier. She made our house a home and couldn't imagine life without her.

Then, as time went on we recognized what a wonderful big sister would be. We decided to expand our family. For most, this is a reasonably simple process but that wasn't our path. We battled the odds and went for IVF. We fell pregnant with our second daughter. As a family we were ecstatic and were so excited when we thought about our life as a family of four.

Unfortunately, our fairy-tale wasn't to be. From our 20 week scan we were seeing specialists and having tests done. I was prodded and poked. I had to have shunts fitted into my womb that would then go into our baby girl's bladder to help drain her urine. The Doctors / Specialists couldn't quite pinpoint what was happening with our baby girl's bladder. They didn't give us much faith... They went on to tell us that the ultra sound scan showed them that our baby girl had Talipes on both feet and that she had two cysts on her brain, a size that they hadn't seen in over 30 odd years! We were asked if we wanted to terminate our pregnancy. This was not an option. We then chose our girls name.... HOPE. She was a fighter and she fought her way to 32 weeks.

Just before was born I had to have a third shunt fitted. Before this took place we were asked if we wanted to terminate our daughter. We were asked this far too many times. Termination was never an option so we asked the Doctors to stop asking us the question as it was never going to happen. She was our and we loved her no matter what.

On 13th August 2013, was born via emergency C section. From the minute was born she brought light and love into our hearts and all those that knew her.

did pass urine, but she never emptied her bladder completely. Dad and I were shown how to catheterize Hope. This had to be done every 4 hours. At a follow-up appointment, we were told we could change what we were doing to 3 times a day. Our girl was heading in the right direction. '---'s little feet were perfect just like her; she never had talipes. We were told that time would tell about the cyst on the brain.

On October 8, 2013 — came home. This was the best day ever, we were a family of four at home. In November 2013, — was admitted into hospital with her 'work' of breathing (Hope was on oxygen 24 hours a day). Two weeks later, — came home. We had our first Christmas at home all together.

In January 2014, was admitted back into our local hospital. Our lives from this day changed. got very poorly and needed to go on a life support machine. was transferred to The John Radcliffe Children's Hospital in Oxford. We spent long periods of time at the hospital. At weekends we would all be together and in the week I would be with always had someone special by her bedside. As a family we were all over the place but we wouldn't have done a single thing differently.

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was diagnosed in January 2014 with the extremely rare lung condition - ACD. None of doctors had come across this condition at The John Radcliffe Hospital.

Doctors found it very hard to answer our question(s) as it was an unknown to them! Doctors were unsure how to treat condition. They would say "---- will tell us what she needs". All we could do was support with her oxygen needs.

An ultra sound was done on stayed the same. Was a clever little soul. She said 'Dad' at 6 months old, then went on and learnt to say 'Mum'. Could stand if she held onto something. She could sit all by herself and she was very proud of herself the first time she did this, and so she should be! She was proving so many doctors wrong.

's pupils never contracted to the light. An eye test was done and 's vision was perfect just like her. At 11 months, had another eye test and again no concerns.

We did have a few days at home, here and there, with and those days were fun family days.

's strength and spirit touched the lives of so many. We believe that Hope is the first child to get to 1 year and 28 days of age with ACD.... We couldn't have been any prouder.

Sadly, our little fighter lost her battle on September 9, 2014 and our hearts are broken. As a family we are still grieving and I don't think we will ever fully recover.

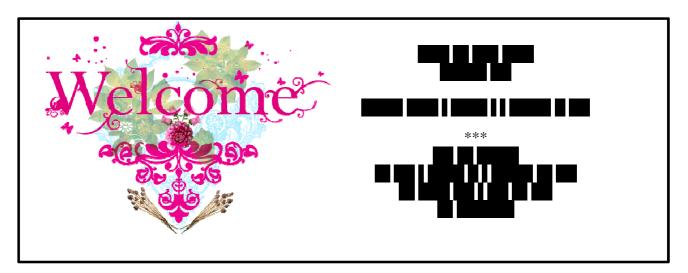
Since losing I have found peace when I run. I put my earphones in and it's me and the road ahead... And my baby girl in my heart. Mile upon mile of time to think, reflect and process what's gone on.

That brings me to where I am now 5 months on. I am training for a half marathon in order to raise money and much needed awareness for the ACD Association (via The David Ashwell Foundation), so that one day no family will ever have to experience the heartache and devastation that we have.

I want my brave baby girl's name to mean something to everyone. I want people to read this and raise awareness and do their bit to bring " into their family's life around the world.







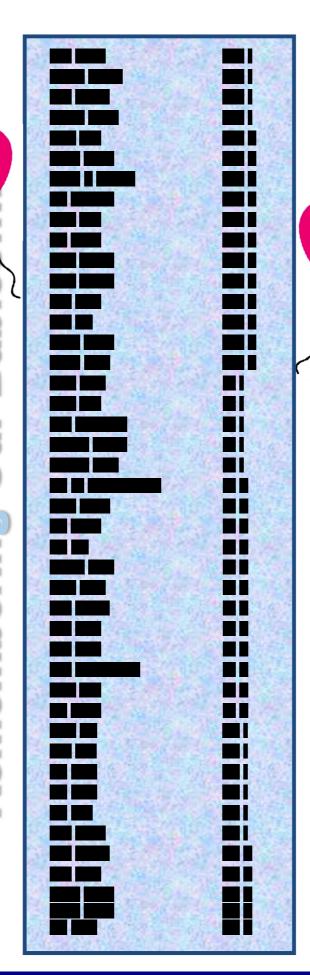
Facebook

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).



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:

 <u>http://givingworks.ebay.com/charity-auctions/charity/alveolar-capillary-dysplasia-association/69889/?favorite=siteadd&favnpid=69889</u>

 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 12 for how to make a donation to NORD.



Thank you to all of you that have made donations to our ACD Restricted Research Fund at			
NORD. The ACDA does not have any way of knowing who you are unless you let us know but			
please know that we are very grateful for your support. A special thanks goes out to			
who made a donation to NORD all the way from Macau SAR (a Special Administrative			
Region of China) in honor of their three children (
honor the work of Professor Pawel Stankiewicz and his team at Baylor.			

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 (which we successfully did again in 2015).

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

Kim Anderson Bush, Secretary - secretary@acd-association.org

Donna Hanson, Treasurer - treasurer@acd-association.org

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http://www.acd-association.org/