

# ACDA NOTES

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Dear Friends and Family,

We are so excited to announce the first major findings on a genetic cause of ACD. Earlier this month, Baylor published an article on their findings identifying a specific gene deletion. This is just the beginning as it points to some related areas for further genetic research. We know that you will be as excited as we are when you read about their findings.

Many thanks to the ACDA members, friends and family members whose contributions have led to this research breakthrough as well as their efforts to raise awareness of ACD.

Fondly,

*Steve & Donna Hanson*

ACDA Executive Directors

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

## Research Update

### Baylor Identifies Microdeletions

There is exciting progress being made in identifying the cause of ACD! Baylor's multi-year research has resulted in findings that were published in the *American Journal of Human Genetics* on June 4, 2009. This publication is the first significant ACD genetics finding and was led by Pawel Stankiewicz, M.D., Ph.D and Dr. Partha Sen, Ph.D at the Baylor College of Medicine in Houston. Below is an abstract of the article. The full article can be purchased at

[www.cell.com/AJHG/abstract/S0002-9297\(09\)00203-1](http://www.cell.com/AJHG/abstract/S0002-9297(09)00203-1)

Abstract: Alveolar capillary dysplasia with misalignment of pulmonary veins (ACD/MPV) is a rare, neonatally lethal developmental disorder of the lung with defining histologic abnormalities typically associated with multiple congenital anomalies (MCA). Using array CGH analysis, we have identified six overlapping microdeletions encompassing the FOX transcription factor gene cluster in chromosome 16q24.1q24.2 in patients with ACD/MPV and MCA. Subsequently, we have identified four different heterozygous mutations (frameshift, nonsense, and no-stop) in the candidate *FOXF1* gene in unrelated

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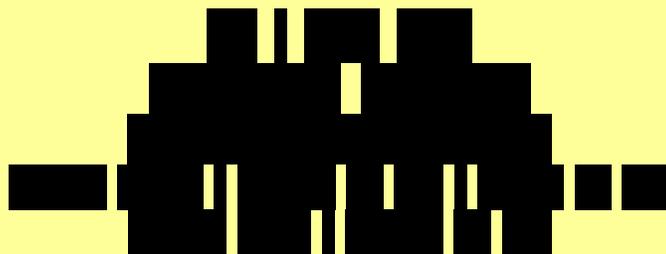
patients with sporadic ACD/MPV and MCA. Custom-designed, high-resolution microarray analysis of additional ACD/MPV samples revealed one microdeletion harboring *FOXF1* and two distinct microdeletions upstream of *FOXF1*, implicating a position effect. DNA sequence analysis revealed that in six of nine deletions, both breakpoints occurred in the portions of *Alu* elements showing eight to 43 base pairs of perfect microhomology, suggesting replication error Microhomology-Mediated Break-Induced Replication (MMBIR)/Fork Stalling and Template Switching (FoSTeS) as a mechanism of their formation. In contrast to the association of point mutations in *FOXF1* with bowel malrotation, microdeletions of *FOXF1* were associated with hypoplastic left heart syndrome and gastrointestinal atresias, probably due to haploinsufficiency for the neighboring *FOXC2* and *FOXL1* genes. These differences reveal the phenotypic consequences of gene alterations in *cis*.

While this article is very technical and difficult to understand, in summary the research has identified some unique genetic characteristics in some ACD samples that occur in a specific area of the genome. The research also infers a mechanism that could explain the genetic anomalies. It is important to note that these genetic changes are unique to ACD samples, but not all ACD samples exhibit them. Nonetheless, this is very important breakthrough on which to focus additional research and is wonderful news for the ACD community. We know that it is just the beginning of finding the answers we all seek. The ACDA thanks Dr. Stankiewicz, Dr. Partha Sen and the many other researchers that contributed to these recent findings.

## Center for Disease Control (CDC) Awareness Campaign

The 3 Angels Memorial Fund for ACD Research is currently working on a budget and plan for Phase 2 of their grant involving production and dissemination of ACD awareness materials. The plan will include finalization of the learning materials - incorporating the genetic findings into the diagnostic protocol, going live with the website, completing a journal article, making a presentation at a national meeting and participation in the Childhood Interstitial Lung Disease (ChILD) virtual case registry.

## New ACDA Family



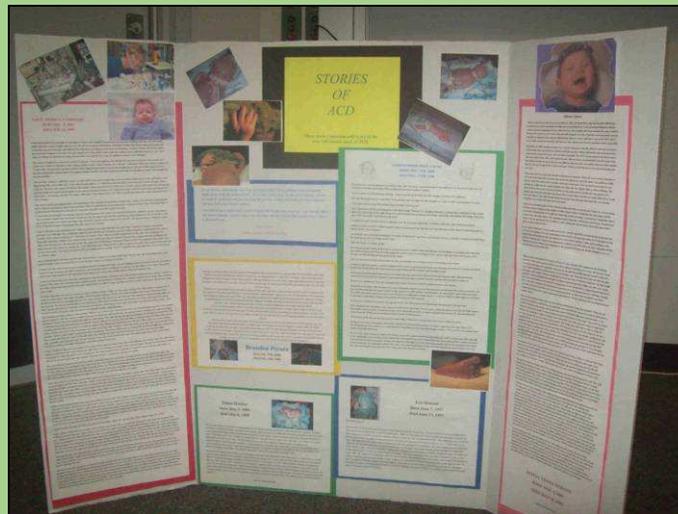
Please contact [redacted] and introduce yourself.

# ACD Presentation Made to Med Students

ACDA member Diana Locke is an operating room nurse at Baptist Medical Center in Jacksonville, Florida. Recently she made a presentation on ACD to 100 second year medical students from the University of Florida, Gainesville. Before her lecture, ACD was not even included or mentioned in the medical students' genetics class which is only three weeks long.

In addition to her presentation, Diana prepared posters that included stories of six babies of ACDA members and medical protocol information that is being defined by the 3 Angels via their CDC grant. Diana said it was "very exhilarating" to be able to tell the story of her son, Christopher, and the other babies that have died of ACD. The students asked a lot of questions and they studied the posters closely as they were being tested on what they learned. Once you look at the photo of one of Diana's presentation boards below, you will realize that our babies' stories and pictures will leave a lasting impression on those students.

So, as Diana said, our stories are being told. Her goal of helping to educate the medical community about ACD is coming to reality. Because of the success of Diana's lecture, ACD will be included in the genetics class at the University of Florida Gainesville every year going forward! She has been invited back next year but she is not resting on her laurels. Her next project is to contact the Florida State University Medical School and the University of Alabama Medical School where she knows more people. Then, on to other medical schools. In addition to her nursing job and raising ACD awareness, Diana also works closely with Wolfson Children's Hospital to plan their annual "Walk to Remember" held every October, and she works with the hospital's bereavement group called "Tear Catchers". She also helps out with bereavement services at the Catholic Church that she belongs to. Thank you, Diana, for telling our stories, increasing awareness in the doctors of the future and making a difference in Christopher's memory.



One of the presentation boards that Diana displayed

# Announcements

- Part of the immunohistochemistry study awarded to Dr. Partha Sen at Baylor by NORD will be performed in the laboratories of Dr. Csaba Galambos. Dr. Galambos is a Pediatric Pathologist at Children's Hospital of Pittsburgh.
- Have keepsake jewelry made of your baby's footprint or toe print at <http://www.memoryinmyheart.com>. Thanks to [REDACTED] [REDACTED] for sharing this keepsake idea.
- If you have recently moved or changed your email address, be sure to let us know. If you have approved release of your information, we share your contact information with new families so they have others to reach out to for support and understanding.
- The donation and acknowledgement process for making a donation to 3 Angels Memorial Fund has been enhanced. Make a general or "In Memory Of" donation for ACD research on the 3 Angels website at:

<http://www.3angelsfund.org/contribute.php>

or by mail:

*Mail check or money order donations to:  
3 Angels Memorial Fund for ACD Research  
PO Box 12251  
Arlington, VA 22219*

## Durand Family Friend Creates Facebook Tribute to Olivia

*Daniela Sainato, a friend of the Durand family in New York, created a Facebook page and moving video (<http://animoto.com/play/beTH2sYfDbIUyjYE75UUog?autostart=true>.) in memory of Lisa and Harry Durand's daughter, Olivia. Lisa said that they never talked about Olivia's passing with Daniela and she was in her early 20's when Olivia passed away. Lisa had no idea that Olivia's death had such an impact on her; however, Daniela created a Facebook tribute in memory of Olivia. Daniela said she questioned her faith when Olivia died because she did not "believe there could be any reason for something like that to happen." But she realized that "you have to live it to the fullest and never take for granted anyone or any minute." The Durands were so moved by Daniela's tribute that they wanted to share it with the ACDA in hopes that others might want to do the same. The link to the Facebook page is:*

<http://apps.facebook.com/causes/282394/20537618?m=65260085>

**Thank you to Daniela for helping us spread the word about ACD!**

# Recent Donations

The 3 Angels Memorial Fund for ACD Research has recently implemented some changes to its donation process. For those of you that are familiar with the NORD process, 3 Angels is setting up a system that is similar to NORD's program. So, when you, a friend or family member make a donation to 3 Angels, the following will happen:

- The donor will receive a thank you note and a tax deductible receipt.
- The parents will receive notification if a friend or family member has made a donation in memory of their baby. This will allow you to say thank you to those that have made a donation.

We are confident that the changes being made at 3 Angels will make the donation process more responsive.

Recently, we have been notified that a few members have made donations to the 3 Angels Fund, and we want to recognize their generosity. Many thanks to the following people:

- First, a very late but very heartfelt thank you to Chris and Donna Hanson who made a generous donation last year in memory of their nephew, Eric Hanson.
- The ██████████ family from New York recently made a large donation from the ██████████ Foundation in memory of their son.
- A friend of the ██████████ family in England recently held a party and accepted donations in memory of ██████████ ██████████ ██████████ ██████████. The funds collected were divided between ACD research and the hospital that cared for ██████████.
- Friends and family members of ██████████ ██████████ ██████████ made donations in memory of their son, ██████████ including:
  - Bonnie & Chris Hatter
  - John Cuttita
  - Ray & Debbie West
  - Town of Halfmoon Sunshine Club (Jo-Ann Smith)
  - Rusty & Debbie West
  - Karrie West & Jeff Hoover
  - Patricia Temple (Town of Halfmoon Co-workers Mindy, Nelson, Pat, Terri, Bonnie, Lisa & Kim)
- Rae and Roger Hanson made a donation in memory of their grandson, Eric Hanson, on what would have been his 12<sup>th</sup> birthday.

If you would like to recognize any family member or friend for their generosity, please contact us. We are so grateful to all of you that have contributed to ACD research.

# Remembering Our Babies

We hope that you will find a special way to celebrate your baby's life.



*Contact Information*

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