ACDA Notes

From the Alveolar Capillary Dysplasia Association

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Dear Families,

Thanks to the efforts of families and doctors around the world, ACDA is becoming a more visible organization. We would specifically like to thank a few people for their recent efforts on behalf of ACD. Madonna Myers has successfully completed the process required for the National Organization of Rare Disorders (NORD) to officially recognize ACD. Dr. Robin Steinhorn, at Chicago Children's Hospital, was instrumental in providing a medical definition for ACD to support this effort. (See the article on Page 2 for more details on these significant milestones). Michele Pezzutti, one of our members in Italy, attended a congress on rare diseases in March and by doing so raised awareness of ACD. His report on the congress is detailed on page 1.

It is through the efforts of people like Madonna, Dr. Steinhorn and Michele that a cure for ACD will eventually be found. We hope that you also will find a way to contribute. ∞

Donna & Steve Hanson

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Searching for Answers, Hoping for a Cure . . .

Italian Convention Held on Rare Respiratory Diseases in Newborns

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Michele Pezzutti, an ACDA father in Italy, attended a convention on rare respiratory diseases chaired by Dr. Marcos Somaschini and held in Bergamo, Italy during March. Dr. Somaschini treated Michele and Patty's daughter, Sara, who he describes in an article published in The Journal of Perinatology in 2000. Due to Dr. Somaschini's interest in neonatal pulmonary disorders, he chaired the *Convention on Rare Respiratory Diseases in Newborns.*

Thanks to Michele's participation, ACD received some visibility in the European and specifically, the Italian medical community. Michele attended the conference session on the "Role of Associations" and summarized that session as follows:

- Europe has an organization called the NEtwork of Public Health Institutions on Rare Diseases (NEPHIRD) in which most European countries participate. Michele will be working with NEPHIRD to ensure that they recognize ACD as a rare disorder.
- There were two suggestions made at the convention on how associations can be most effective:
  1) There is power in numbers. Rare diseases that are similar in nature or require similar research can be more effective when grouped together into an "umbrella" organization. It is also beneficial to have a large number of members so our voice can be heard.
  2) Identify one single point of contact with the medical community and other government institutions, instead of having a lot of fragmented contacts with a lot of interfaces.
- ACDA was mentioned by one of the presenters during her speech regarding the role of associations. She cited ACDA as an example of an association that is easy to find and to reach, thanks to its internet site. Congratulations to Madonna and Phil Tenney for the website!!!

Many thanks to Michele for attending this convention, providing a summary to keep all ACDA members informed, and for his continuing effort to gain exposure for ACD in the European community!! ∞

Recent ACDA Accomplishments

• NORD Recognizes ACD

Thanks to the efforts of Madonna Myers, the founder of ACDA, the United State's National Organization of Rare Disorders (NORD) now recognizes ACD as an orphan disease. As of this writing, the NORD website lists the ACDA, but does not yet list ACD. We will let you know when it does, or you can check their website periodically. Most importantly, this means that anyone can now contribute money to NORD and earmark the funds for ACD research. A minimum of $35,000 is needed for NORD to fund a grant to study ACD so please be sure to let friends and family know that they can now help us make an impact.

You can contribute to NORD for ACD research by sending a check to the following address**. Please make your check payable to "ACD Fund of NORD" to earmark your donation for ACD research**. If you have any questions, please contact Madonna Myers.

The National Organization for Rare Disorders, Inc.
P.O. Box 8923
New Fairfield, CT 06812-8923
(800) 999-6673 or (203) 746-6518
Fax: (203) 746-6481
http://www.rarediseases.org

Matching Gifts:
If your employer has a Matching Gifts Program for charitable organizations, your contributions to NORD can grow! Please check with your Human Resources Department.

• Medical Definition for ACD

NORD received a grant from the National Institute of Health to publish a doctor's reference guide for rare diseases. A definition for ACD was required for NORD to add it to the reference book. Many thanks to Dr. Robin Steinhorn, a Professor of Pediatrics, the Head of the Division of Neonatology and an Associate Chair of Pediatrics at the Children's Memorial Hospital in Chicago and Northwestern University, who authored the medical definition for ACD. Not only has Dr. Steinhorn treated ACD cases and authored articles on ACD, she has been active in the ECMO community and has been instrumental in providing guidance to the ACDA. Dr. Steinhorn's definition of ACD that will be included in NORD's rare disease book is included below. We know everyone joins us in sending a special thanks to Dr. Steinhorn for her contribution and all her work on behalf of ACD.

Alveolar Capillary Dysplasia

DEFINITION: Alveolar capillary dysplasia (ACD) is a lethal developmental anomaly of the pulmonary vasculature: it is generally described as the failure of formation of the normal air-blood diffusion barrier in the newborn lung. Alveolar capillary dysplasia is usually associated with “misalignment” of the pulmonary veins.

Synonyms: Misalignment of pulmonary veins

DIFFERENTIAL DIAGNOSIS: Idiopathic persistent pulmonary hypertension of the newborn, surfactant protein B deficiency

SIGNS & SYMPTOMS: Alveolar capillary dysplasia is a pulmonary disease that presents in very early infancy. Infants generally become critically ill in the first days of life with severe hypoxemia and pulmonary hypertension, although presentation has been reported at 6 weeks of life in an infant with a patchy distribution of disease. The majority of patients with ACD will have other associated anomalies of the cardiovascular, gastroinstestinal, urogenital, or musculoskeletal systems.

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The initial presentation is identical to severe idiopathic pulmonary hypertension of the newborn. However, infants with alveolar capillary dysplasia do not respond, or respond only transiently to therapies that are usually effective in reversing this condition. Infants with ACD do not improve despite maximal support in the intensive care nursery including mechanical ventilation, nitric oxide, and extracorporeal membrane oxygenation (ECMO).

ETIOLOGY/EPIDEMIOLOGY: Fewer than 50 cases of alveolar capillary dysplasia have been reported in the literature. No sex predilection has been identified. While the cause is unknown, there have been six cases reported in siblings, indicating that in some cases this may be a familial disorder with autosomal recessive inheritance.

DIAGNOSIS: The diagnosis of alveolar capillary dysplasia should be considered in infants who present with severe hypoxemia and idiopathic pulmonary hypertension, and who do not respond appropriately after 7 to 10 days of neonatal intensive care treatment as described below. The majority of patients with ACD (approximately 75%) will have other associated anomalies of the cardiovascular, gastrointestinal, urogenital, or musculoskeletal systems. The initial chest radiograph is usually normal. If a cardiac catheterization is performed, there may be absence of the capillary blush phase.

The diagnosis can only be confirmed by lung biopsy or autopsy. Consulting a pathologist with experience in making this diagnosis may be helpful. Pathological features include a paucity of alveolar capillaries, widened alveolar septae, and increased muscularization of pulmonary arterioles. There is usually malpositioning ("misalignment") of pulmonary veins in the bronchovascular bundle, but this is not required for the diagnosis. A focal distribution of disease has been described, which makes it necessary to examine multiple lung sections if ACD is suspected.

TREATMENT:

Standard Therapy: Standard therapies include mechanical ventilation, high concentrations of inspired oxygen, inhalational nitric oxide and extracorporeal membrane oxygenation (ECMO) support. These therapies prolong life by days to weeks, but have not led to long-term survival. The longest reported survival is to 2 months of age with the use of extracorporeal membrane support followed by inhaled nitric oxide.

Investigational Therapy: Theoretically, alveolar capillary dysplasia could be treated by lung transplantation. However, successful transplantation has not yet been reported. Donor availability continues to limit the utilization of lung transplantation for neonatal diseases. ∞

- Please be sure to forward any change of status information to us including new address, phone number and e-mail address. If you want to be removed from the association at any time, please let us know. We don't want to intrude.

- The National Human Genome Research Institute (NHGRI) and the National Institutes of Health's Office of Rare Diseases (ORD) in the United States have created an information center that can provide free and immediate access to accurate, reliable information about genetic and rare diseases to patients and their families. We are pleased to announce they were able to provide information to us on ACD. To receive ACD information, send them an e-mail at GARDinfo@nih.gov
- This newsletter is open to things you would find helpful. Please feel free to send us ideas for stories, poems, websites and support groups you found helpful, and pictures and stories of your baby.
- Please remember that the ACD study team from Baylor is interested in talking to all families that have lost a baby to ACD. If you are interested in receiving more information, please contact Dr. Bassem Bejjani at (713) 798-5061. ∞

Safe Arrivals

Congratulations to the following families who have been blessed with another baby:

[redacted]

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[redacted]

Remembering

Go ahead and mention my child
The one that died, you know
Don't worry about hurting me further
The depth of my pain doesn't show.
Don't worry about making me cry
I'm already crying inside
Help me to heal by releasing
The tears that I try to hide.
I'm hurt when you just keep silent
Pretending it doesn't exist
I'd rather you'd mention my child
Knowing that he has been missed.
You asked me how I'm doing I say
"pretty good" or "fine"
But healing is something on-going
I feel it will take a lifetime.

By Elizabeth Dent

Please Welcome Our New Families . . .

It is with mixed emotions that we'd like to introduce the families that have joined ACDA since the last newsletter. Please take the time to introduce yourself, offer support and share the story of your child (children). You may find a family in your area.

[redacted]

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[redacted]

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[redacted]

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[redacted]

UPDATE:
The Wagner family recently contacted ACDA with the news that their daughter's final autopsy report revealed she had congenital cystic adenomatoid malformation (CCAM), not ACD. CCAM is a disease where the bronchial alveolar spaces are not connected. They have requested to be removed from the ACDA. We wish them well.

Remembering your baby!...

Memorials and mementos help us express our feelings and also honor our baby's importance. Here are two suggestions that you might consider to memorialize your child:

- Plant a tree in your yard to give you something meaningful to nurture;
- Have your baby's name engraved in stone or brass and place it somewhere as a special remembrance.

If you have found a unique way to celebrate your child's life and would like to share it with the ACDA, please e-mail us at sdhanson@flash.net. ∞