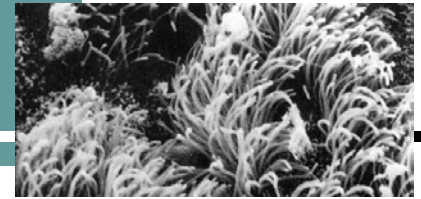


# PCD NEWS



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## Upcoming Events

PCD Foundation  
National Walk-a-Thon  
October 10, 2004

Gordon Research Conference on Cilia, Mucus and Mucociliary Interactions  
February 27-March 4, 2005, Rancho Santa Barbara, California

American Thoracic Society International Conference  
May 20-25, 2005, San Diego, California

## WHAT'S IN A NAME?

Kartagener('s) syndrome, immotile cilia syndrome, dyskinetic cilia syndrome, primary ciliary dyskinesia--the condition is difficult enough to explain without having to wade through a maze of competing names! Here's a simple (sort of) explanation of how ciliary disorders got into this naming predicament, and why they defy common nomenclature to this day.

To fully understand the problem we have to have some historical perspective. Western medicine has a long-standing tradition of naming diseases after the physicians who identified them. This is known as eponymous" (ee-pon-a-mus) naming, and some common examples include Down('s) syndrome, and Alzheimer('s) disease. Not surprisingly, serious debate frequently ensued over the assignment of eponymous credit, because it was generally the first person to *publish* findings who received credit, not necessarily the first person to *identify* a set of symptoms as a disease entity.

In recent years, there has been a movement to eliminate eponymous names. There are several valid reasons for this: 1.) it is frequently impossible to know who actually deserves the credit for disease discovery, 2.) as the underlying pathology for diseases becomes known, it is more accurate for disease names to reflect that pathology, and 3.) there tends to be a strong Western bias to eponymous naming that may ignore the contributions of other cultures.

The first step to reducing the use of eponymous names was to remove the possessive apostrophe "s." This was done in an effort to clarify that specific diseases do not "belong" to any particular person. Thus, Down's syndrome is now written as Down syndrome and Kartagener's syndrome is written as Kartagener syndrome. (By the way, only eponymous disease names and Latin genus names are capitalized. Other disease names, such as cystic fibrosis and primary ciliary dyskinesia, should not be capitalized)<sup>1</sup>.

Sticklers would like to see eponymous names go the way of the dinosaur. While it is hard to imagine this ever happening, they have some very compelling arguments. Take, for instance, Kartagener syndrome (KS). KS is named for a Swiss physician, Manes Kartagener who, back in 1933, noticed a pattern of symptoms that included bronchiectasis, situs inversus, and otitis media in a number of his patients. The technology to understand the pathological process leading to these seemingly unrelated symptoms was not available to Dr. Kartagener, so he was forced to publish his findings without identifying an underlying cause.<sup>2</sup> As observant as Dr. Kartagener was, he was not actually the first physician to publish findings related to this triad. This honor goes to a Russian doctor, AK Siewert, who recognized the distinct features of KS back in 1904! So would it be more accurate to call Kartagener syndrome "Siewert syndrome" (some do) or perhaps "Siewert-Kartagener syndrome?"

CONTINUED NEXT PAGE

## WHAT'S IN A NAME? (CONT.)

The preferred naming method today is to indicate the pathology in the disease name

Wait a minute! There's another really important eponymous guy in this story. Bjorn Afzelius, a Swiss researcher, noticed that a number of males with immotile sperm also had chronic respiratory problems and otitis media. Some of them (roughly 50%) also had situs inversus, which, he surmised, was a consequence of the underlying ciliary dysfunction and not a disease process in itself. It was Dr. Afzelius who actually proposed that ciliary dysfunction was responsible for all three problems and in 1976 coined the term "immotile cilia syndrome" to describe his findings. So then would it be more appropriate to call it "Siewert-Kartagener-Afzelius syndrome?" Obviously this could quickly get out of hand.

The preferred naming method today is to indicate the pathology in the disease name. Thus trisomy 21 (literally 3 copies of chromosome 21) is preferred to Down syndrome because it clearly describes the cause of the symptoms, and primary ciliary dyskinesia with or without situs inversus is preferred to Kartagener syndrome or immotile cilia syndrome because it reflects the nature of the underlying defect. In fact, European doctors have pretty much standardized their nomenclature to PCD or PCD/SI (for patients with situs inversus).

Many of us have been told that we (or our children) have "immotile cilia syndrome," a term that certainly describes a particular defect, but is considered obsolete by most clinicians and researchers familiar with ciliary disorders. The reason for this is that it does not indicate the genetic nature of the disorder (cilia may be rendered immotile due to secondary causes), and because improvements in technology have shown that it is rare, although not impossible, for cilia to be truly immotile. Most researchers involved in assessing ciliary slides for motility confirm that evaluation using newer technology will almost always show some—albeit severely dysfunctional—ciliary motion.

The research/medical community has adopted the term primary ciliary dyskinesia for this group of disorders and it is likely to stick for the near future. Because supporting research efforts is a big part of the mission of the PCD Foundation, we have also adopted this naming preference. Since raising awareness of this disorder is one of our primary objectives, we are optimistic that identifying the disorder by a simple acronym like PCD will help us to achieve this goal. However, the important point is that, whatever you choose to call it, Kartagener syndrome, immotile cilia syndrome, and ciliary dyskinesia are ALL PART OF THE SAME DISEASE PROCESS—at least as far as we know right now. There are multiple gene mutations involved in ciliary disorders, but the outcome of these defects is the same—the cilia don't do their job correctly—resulting in chronic lung, sinus, and ear infections, and random (50/50) organ orientation.

<sup>1</sup>American Medical Association Manual of Style, 9th Edition; pp 469-70.

<sup>2</sup>To truly qualify for a diagnosis of Kartagener syndrome, a patient *must* have all three symptoms. Thus, a young child with situs inversus, chronic upper and lower respiratory illness, and otitis media won't technically "qualify" as a Kartagener syndrome patient *until* bronchiectasis develops. Of course, this is ridiculous—but it HAS actually happened! It is unlikely that Dr. Kartagener intended for such strict adherence to the "triad" of symptoms, but it highlights the problem with syndromic diagnosis—the clinician is limited to looking for obvious symptoms because the underlying pathology is not understood.

## NIH AND THE OFFICE OF RARE DISEASES APPROPRIATE FUNDS FOR PCD RESEARCH!!!

There are more than 6,000 known rare diseases affecting over 25,000,000 people in the U.S. Recognizing a large unmet need for clinical research in rare diseases, Congress mandated the creation of an Office of Rare Diseases (ORD) in 2001. ORD, a division of the National Institutes of Health, was faced with a daunting task—how to divvy up limited resources for such a diverse group of diseases. The solution they adopted was to develop a network, the Rare Diseases Clinical Research Network (RDCRN). The goal of the RDCRN is to provide access to research to a large number of rare diseases by encouraging groups with similar conditions and research goals to work together. Under this arrangement, ORD provides funding for disease groups, rather than for individual diseases.

The Genetic Diseases of Mucociliary Clearance consortium is the newest research consortium to be funded by the RDCRN. The principal investigator (PI) for this consortium is Michael Knowles, MD from the University of North Carolina, Chapel Hill. Dr. Knowles' grant proposes the development of a network of clinical sites with the expertise to diagnose and treat PCD, and to compare clinical outcomes and disease progression in PCD to atypical cystic fibrosis, pseudohypoaldosteronism (PHA), and other rare conditions that impair mucociliary function. Co-investigators for this grant are Margaret Leigh, MD, **University of North Carolina, Chapel Hill**; Tom Ferkol, MD and colleagues from **Washington University, St. Louis**; Ronald Gibson, MD and colleagues from the **University of Washington, Seattle**; and Scott Sagel, MD and colleagues from the **University of Colorado, Denver Children's Hospital**. These four sites will serve as research "hubs" and will be equipped and trained to fulfill the requirements of the research study. In addition to the four research hubs, there are currently 12 participating academic sites in the United States, Canada, and Puerto Rico

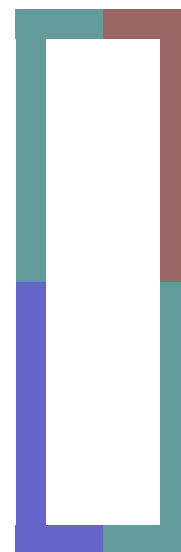
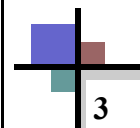
that will identify potential research candidates and follow patients clinically. In addition to funding research, the RDCRN is partnering with patient groups to make information about rare diseases more accessible to patients and the general public. This effort includes a number of initiatives: a RDCRN website ([www.rarediseasesnetwork.org](http://www.rarediseasesnetwork.org)), a working committee of representatives from the patient advocacy community who will meet with the research steering committee to represent patient interests, and a commitment to help groups develop "best practice" guidelines for continued success in their growth.

The ORD and the RDCRN are committed to rare disease research and to supporting the efforts of patient advocacy groups. For more information, contact the ORD at <http://rarediseases.info.nih.gov/>. Information and updates will also be posted to the PCD Foundation website ([www.pcdfoundation.org](http://www.pcdfoundation.org)) after September 15.

### A SPECIAL GIFT

WHEN STRANGERS GATHER,  
FRIENDSHIPS GATHER,  
WORDS OF KINDNESS GATHER.  
WE ARE ALL HERE FOR A REASON  
FOR SUPPORT OF ONE ANOTHER!  
STORIES OF SORROW AND JOY  
CAN BE NO MORE,  
WITH THE SUPPORT OF EACH OTHER!  
TRAVEL IS NO OPTION EITHER – WHETHER  
IT BE GENTLY PLACING MY FINGERS ON A  
KEYBOARD  
OR BY COMING BY PLANE --  
WE ARE ALL HERE FOR EACH OTHER  
AND  
IF WISHES WERE KNOWN  
MORE FRIENDSHIPS WILL BE FOUND!  
ON TOP OF BUILDINGS, IN DOCTOR'S OFFICES,  
IN PCD/KS FORUMS,  
AND  
**ON PCD FAMILY DAY.**

Contributed by Betty (PCD patient, AZ) in honor of PCD Family Education Day.



# Foundation News: It's Official...

We are now officially a 501(c)(3) tax-exempt organization! Our tax-exempt status was granted retroactively back to November 2003. Prior to that date, we were operating as a program of the Vest Foundation, and they were technically our "fiscal agent." This worked well as a way to get started, and the Vest Foundation was our chief source of financial support in the early years. Times change, however, and corporations are bought and sold, and the Vest Foundation is now under new management. It was an ideal time to strike out on our own. Secretary of the Board and attorney, Eric Lefkowitz, filed the required paperwork and we were notified of the approval in mid-April.

What does this mean for us as a patient organization? Being a tax-exempt patient organization lends credibility to our fundraising and grant-seeking activities. It also gives us control over our finances and allows us to establish our own financial priorities. Of course, there are additional responsibilities with our new status, as well, including IRS reporting requirements and the need to develop our own operational budget. The grid below compares the operating budget and staffing of three other genetic lung diseases. The CF Foundation is, of course, phenomenally successful and has been doing this for decades. At one point, however, they were just a small group of concerned parents and patients, like us, who managed to develop an organization that is

one of the premiere research-funding bodies in the world. LAM and Alpha-1 have not been organized for as long as CF, but they have accomplished tremendous things in a short amount of time, and are a great source of information for us.

The PCD Foundation Board is working on Foundation-based fund-raising strategies, including a national PCD Walk-a-thon (see Gina Manning article, page ) and a sponsorship program (modeled after a successful LAM Foundation program they were generously willing to share with us). The goal is to set up an endowment fund—money that is set aside to earn interest, which is then used to cover operating expenses—so that all future fund-raising activities can be research and education focused. The fund-raising plan will be announced at PCD Family Education Weekend in July and will be posted to the website at that time. Keep sending in your ideas! And feel free to initiate fundraising activities of your own (e.g. garage sales, bike-a-thons, special events). With our new status as an independent organization, the recognition of PCD as a disease of scientific interest to the National Institutes of Health, and the continued development of technologies to diagnose and treat ciliary disorders, momentum for moving forward with this organization has never been higher!

## Genetic Lung Disease Patient Organizations\*

Organization:	Number of Patients (U.S.):	Paid Staff:	Operational Budget 2003:
CF Foundation	~30,000	100's	Revenue: 75,030,729* Assets: 162,889,933*
Alpha-1 Foundation	-100,000 severe (estimated) -25,000,000 total (estimated) -Fewer than 10,000 currently diagnosed	22	Revenue: 5,347,911* Assets: 2,221,991*
LAM Foundation	~1,000 (estimated)	3	Revenue: 696,025* Assets: 595,162*
PCD Foundation	~20,000 (estimated) -No current statistics on number actually diagnosed	0	Revenue: 1,000 Assets: 0

Where do we stand? Here is current financial information comparing the PCD Foundation to groups with similar patient populations and goals. This information is provided to highlight our challenges as a developing patient group and to identify benchmarks and goals we can set for ourselves.

\*Financial information from Guidestar.com, available online



## PCD Foundation National Walk-A-Thon

Mark your calendars for October 10, 2004—the date of the first-annual PCD Foundation National Walk-a-Thon! After a successful pilot walk (see Gina Manning's experience below), we decided to expand the event nationally. Packets for walk organizers will be available from the PCD Foundation starting August 16. Gina Manning has agreed to organize the walk effort. For more information, please contact Gina (manning\_gina@yahoo.com) or Lynn Ehrne (NEPCDchapter@frontiernet.net).

### Organizing a Walk—My Experience by Gina Manning

I was thinking of having a small walk to raise funds for family day. My niece, Amber, was a big help in recruiting walkers. I work for the Hartford Public Schools and know the kids and staff. This made it easy to get their help. The school groups of students against destructive decisions (SADD), the National Honor Society and the student council were willing participants. My family was a support system as well.

I first asked my family to walk and get sponsors. Amber passed out sponsor envelopes to the high school students. I gathered donations from co-workers, school staff members, healthcare providers, local businesses, family and friends. I did this by one-on-one contact and mailing out around 100 letters to individuals and businesses. I also contacted local newspapers and radio stations to announce my walk-a-thon. One local newspaper ran a small article about the walk-a-thon.

We raised and collected over \$2200.00 in donations for the foundation. We had around twenty walkers come out. Due to the rain we started in the middle school gym but finished outside. We walked over an hour and had a great time!

Having the walk not only raised funds for the PCD Foundation, but it informed others about

### Gina (continued)

PCD and what it is. Because of the walk, many people in my area are now aware of PCD. I also hope to create a local support group to bring PCD patients together to raise more funds for the PCD Foundation and to support one another.

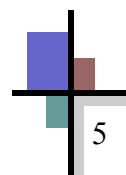
We're looking for even bigger and better things next year at our second annual PCD Foundation walk-a-thon!

### PCD Foundation Website Upgrade

The PCD Foundation website has a new look! For now, there have been minimal changes to content, but we are in the process of phasing in a more sophisticated website with additional capabilities. Proposed upgrades to the site include: 1.) a site-specific search engine, 2.) re-organized categories for accessing information, 3.) an area for healthcare professionals to get research and treatment information, 4.) video clips of common diagnostic procedures (nasal biopsy, NO testing, etc) and of common treatments for PCD patients (HFCWO, inhaler and SinuNeb technique, etc), 5.) online donation capability, 6.) marketing materials that can be downloaded and printed at home, and 7.) a PCD Foundation sponsored message board.

The Internet is an excellent tool for accomplishing many of the major goals of the PCD Foundation, e.g. education, outreach, and patient support. We have excellent people working with us to upgrade our site, and will implement changes as time and finances allow. Getting the site moved to its new hosting service was a major step in this process. Your feedback will help us to insure the PCD Foundation website is meeting the needs of the PCD patient/family community.

[www.pcdfoundation.org](http://www.pcdfoundation.org)



## PCD Foundation

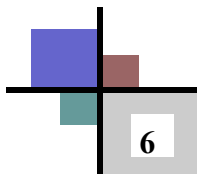
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Email: [info@pcdfoundation.org](mailto:info@pcdfoundation.org)

We're on the Web!!!

[www.pcdfoundation.org](http://www.pcdfoundation.org)

*Education & Advocacy for  
People with Inherited Ciliary  
Disorders*



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## Journal Watch NEW ARTICLES OF INTEREST TO THE PCD COMMUNITY

The National Library of Medicine (NLM) maintains a database of peer-reviewed articles from international medical journals. The peer-review process means that the article is subject to vigorous critical review by a panel of experts prior to being accepted for publication. This information can be accessed by the public at: <http://www.ncbi.nlm.nih.gov/PubMed> or by typing "entrez pubmed" into your internet provider search function.

When you enter a query (e.g. primary ciliary dyskinesia) into PubMed, a list of articles will appear, most recent at the top. Click on each article for a link to its **abstract**, or brief overview. Often, the abstract provides enough information and there is no need to get the full article. If the full article is desired, there are several ways to obtain it; 1.) universities with medical schools frequently maintain a large inventory of medical journals. Copies of articles from these journals are usually available to the public for a small fee, 2.) articles can be ordered from the publisher for a (typically exorbitant) fee, and 3.) the PCD Foundation maintains a bibliography of many articles and may be able to provide a copy.

- ◆ Chodhari R. Cilia, primary ciliary dyskinesia and molecular genetics. *Paediatric Respiratory Review* March, 2004, volume 5; issue 1, pgs 69-76.
- ◆ O'Callaghan C. Innate pulmonary immunity: cilia. *Pediatric Pulmonology Supplement* 2004, volume 26, pgs 72-73.
- ◆ Coste A. Atypical sinusitis in adults must lead to looking for cystic fibrosis and primary ciliary dyskinesia. *Laryngoscope* May 2004, volume 114; issue 5, pgs 839-843.