

THE BEAT



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Did You Know...

• Participation in PCD Foundation committees is open to everyone. The committees will be working on a number of projects in 2006 (e.g. insurance coding for PCD, transplant issues in PCD, improving our web presence, etc.). If you are interested, please call the foundation at 612-965-3116 or at www.pcdfoundation.org.

• The Fourth Annual PCD Family Education Day will be held at the Millennium Hotel in Buffalo, NY August 12, 2006

HAPPY 2006!!!

2006 is shaping up to be a very busy year for the PCD Foundation, but before we focus on the future, let's take a few minutes to look at what we accomplished in 2005.

This year we continued to build awareness about PCD within the research community by participating in the American Thoracic Society (ATS) meeting and the Rare Lung Diseases Consortium meeting. In addition to clinical presentations about PCD, we were able at both meetings to present the patient perspective at both events with speakers from the PCD community.

The third-annual PCD Family Education Day was held in St. Louis in 2005. Family Day continues to be our premiere patient education event and it is always a pleasure to meet with families and have the opportunity to socialize and learn about PCD. This year's event was held in conjunction with a PCD research meeting, so participants had the chance to meet some of the researchers and put faces with the names we hear so often.



The PCD Foundation Board nearly doubled in size in 2005. With the additional help and an infusion of enthusiasm, we were able to create committees focused on specific priorities, including:

- Identifying and prioritizing research opportunities to be prepared to award small PCD Foundation research grants in the future (Research Committee).
- Developing printed and web-based educational materials for a variety of audiences (Education Committee).
- Assessing and addressing the needs of PCD patients dealing with end-stage lung disease issues (Adult Care Committee).
- Managing the organizational and legal needs of the foundation (Executive Committee).
- Maintaining a revenue stream (Fundraising Committee) to make all of the above possible.

Looking Ahead to 2006

2006 is going to be an important year for PCD. We anticipate successful navigation of some of the bureaucratic hurdles that have delayed the Rare Diseases Clinical Research Network study allowing the study to begin enrolling patients. Once this happens, the network of clinical sites with expertise in treating PCD will rapidly come on line. We can look for continued progress in other research areas, as well: identifying additional PCD causing genes, developing diagnostic tests to look for known mutations, and continuing efforts in understanding the nitric oxide connection and other mysteries of PCD.

PCD Family Education Day—One Family's Experience

by Carolyn Schroeder

This past July, my husband Brian and I packed up our two kids and drove from our home in suburban Chicago to St. Louis. We were on our way to attend our first ever PCD Family Day. Our son, Liam, was diagnosed with PCD in October of 2004, when he was two years old. We had some initial panic at being diagnosed. Even our wonderful ENT had admitted she knew very little about the condition and the first pulmonologist we saw told us he had to do some research on it to answer our questions. Fortunately, I found the PCD Foundation website and Michele Manion took the time to speak with me personally about how to raise a child with this condition. I am very grateful she took the time to talk to me and tell me about her own experiences with her adult daughter. She gave me hope at a point where I was simply overwhelmed and enabled me to see a future for Liam.

When we saw the posting about PCD Family Day, we were excited at the prospect of being able to speak directly with the various doctors we had been told were concentrating on PCD. We were even more excited about meeting other people who are living with PCD in their families. Family Day turned out to be a great experience on several different levels for our family!

When we arrived in St. Louis, we were able to meet Dr. Johnny Carson, the very doctor who had seen Liam's biopsy and made his diagnosis. We were able to discuss PCD with him personally and ask several questions which caused us concern. He was able to tell us of other cases of PCD where, once on a treatment plan, the child was able to conduct a fairly normal life. He explained the difficulty in formulating an opinion on a biopsy and explained why he likes to see the patient himself and prefers to take the biopsy himself because of the difficulty in obtaining sample from a complicated three dimensional structure. My husband was fascinated by this discussion.

The next day we headed off to the St. Louis Zoo where the Foundation had arranged for a conference room. This was a perfect location for a family with kids! My husband and I were able to switch out taking the kids around the zoo and listening to the various speakers. We learned so much!

Every aspect of living with PCD was covered. The clinical or research side was explained by Dr. Carson with visual aids. It is so difficult to explain what it really means to be missing inner dynien arms but he was able to do so. We heard from Dr. Margaret Leigh about infection control and steps we can take to minimize the risk to Liam. Dr. James Forsen spoke about ear and sinus involvement in PCD and we were surprised to hear him report that there was little evidence of ear tubes being beneficial to the PCD patient. I went up and spoke to him after his presentation, one of the fabulous things about Family Day was the accessibility and the approachability of all the speakers. I told him that our experience with ear tubes was just the opposite. They have been hugely beneficial to Liam and made his continuous ear infections largely a thing of the past. Dr. Forsen, my husband and I, were able to discuss at length the need for research on the management of the condition. I took this information back to our ENT and she and Dr. Forsen are corresponding about sharing information.

Aside from the medical aspect of Family Day, the best benefit to me, as a parent, was meeting other people who are coping, to varying degrees, with the same issues we are, day to day. We met a mother who has two PCD kids, both adults and she gracefully shared her experiences as a mom raising kids (without a diagnosis until their twenties!) with issues nobody else understood. We heard from a teenager with PCD and what his experience is like, going through adolescence with this condition. We also heard from his parents. Knowing that this will be us someday, I am grateful they told their stories. Several of the parents spoke about how they handle the issues relating to attending school. How they let school friends and others know about the condition. I know I will use their insights on various ways to deliver information on PCD when Liam starts school. There was also a speaker on the emotional aspects of living with this condition. I found this very helpful. Because PCD is genetic, I have often felt guilty that I caused this. Just having a roomful of people having similar experiences was wonderfully supportive. Best of all, my boys made a friend with a girl who has PCD. Although she is a lot older than they are, they spent most of the day playing with her and exploring the zoo.



Liam Schroeder, Aiyana Britton & James Schroeder

Going to PCD Family Day was a wonderfully positive experience for our whole family. My husband and I were able to learn a great deal, both medically and socially how to manage our lives with PCD. We have always viewed this as a family effort and Family Day simply reinforced that idea. Not only the lack of information, but the lack of correct information is probably the greatest obstacle to the newly diagnosed. As a community we are really here to help each other through research and sharing information, even the basic information that the newly diagnosed might not know. PCD Family Day is just the type of forum that enables this group to maximize our assets, our life experiences. Towards the end of the day we spoke about raising money for the Foundation in order to seed research efforts. My husband and I recently hosted a walk to benefit the Foundation. It is our way of helping an organization that has really lent a helping hand to us, and we plan of this being the first of many events to come!

ASK THE EXPERTS

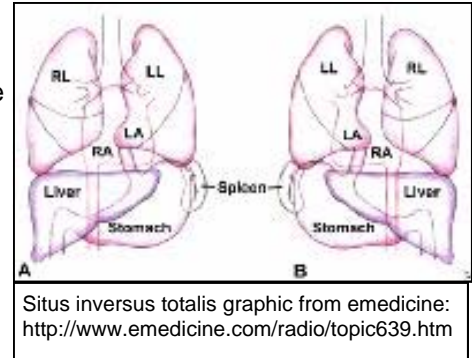
Primary Ciliary Dyskinesia and Kartagener's Syndrome: What's the Difference?

Marcus Kennedy MD and Michael Knowles MD
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The vast majority of the human population has identical location of their organs in the chest and abdomen. Specifically, the organs are asymmetrical with the liver on the right; the heart, stomach and spleen on the left; and the left lung had two lobes and right lung has three lobes. This "normal" positioning of internal organs is called *situs solitus*.

Abnormalities of this normal organ location occur in 1/5000 people and fall into two categories: 1) *situs inversus totalis* and 2) *situs ambiguus* (also known as heterotaxia). *Situs inversus totalis* refers to a complete mirror image reversal of the chest and abdominal organs, with the liver on the left; heart, stomach and spleen on the right; and the lungs are reversed (3 lobes on the left side). The prevalence of *situs inversus totalis* is 1/15,000.

Primary ciliary dyskinesia (PCD) is characterized by chronic cough, chronic rhinitis, and chronic sinusitis. The underlying cause is a defect of cilia in the airways, making them unable to beat (ciliary immotility- hence the old name "immotile cilia syndrome") or unable to beat normally (ciliary dyskinesia). Approximately half of patients with PCD have normal positioning of their organs (*situs solitus*). The other half of PCD patients have their organs reversed (*situs inversus*), which is part of the "triad" that Kartagener described in 1933 (Kartagener's syndrome). This triad consists of chronic bronchiectasis, chronic sinusitis and *situs inversus*. Therefore, approximately half patients with PCD have Kartagener's syndrome, whereas all Kartagener's syndrome patients have PCD.



It is now known that PCD patients develop *situs solitus* or *situs inversus* as random chance. For example, some siblings and twins with PCD (with the same genetic and ciliary structure defect) display different *situs* types, one sibling with *situs solitus* and the other with *situs inversus*. The severity of symptoms and disease appears to be the same in PCD patients with or without *situs inversus*.

Why do some patients with PCD have *situs inversus* and therefore Kartagener's syndrome? The development of organ lateralization is a complex process which begins early during fetal development. In an area of the developing fetus called the embryonic node, ciliary function partially controls organ lateralization. If these cilia are not working properly, *situs* abnormalities develop in approximately 50% of births.

A small number of patients with PCD have *situs ambiguus* which refers to patients who do not fall into either the *situs solitus* or *situs inversus* categories. An example is patients with reversal of abdominal organs only.

In conclusion, organ location is dependant on functioning cilia. Ciliary dysfunction in PCD leads to abnormalities in organ location and lateralization in approximately 50% of individuals. The majority of these patients appear to have *situs inversus totalis* (and therefore what is currently called Kartagener's syndrome). A minority appear to have *situs ambiguus*. Severity of symptoms and disease is not affected by *situs* type.

Journal Watch—Recent Peer-Reviewed Articles of Interest to the PCD Community

Primary Ciliary Dyskinesia: Clinical Presentation, Diagnosis and Genetics. KS Van Gravesande and Heymut Omran. *Annals of Medicine* 2005;37(6):439-49 .

RPGR is Mutated in Patients with a Complex X-linked Phenotype Combining Primary Ciliary Dyskinesia and Retinitis Pigmentosa. A. Moore, Estelle Escudier, et al. *Journal of Medical Genetics* July 31, 2005.

Correlation Between Cough Frequency and Airway Inflammation in Children with Primary Ciliary Dyskinesia. N. Zihlif, E. Paraskakis, C. Lex, L. Van De Pohl, and Andrew Bush. *Pediatric Pulmonology* 2005 Jun;39(6):551-7.

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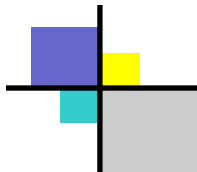
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*Education & Advocacy for
People with Inherited Ciliary
Disorders*



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This publication is provided for general information only and is not intended to replace the advice of your medical professional. Please consult your physician before making any changes to your treatment regimen.

Looking Ahead to 2006

Continued from page 1.

Thomas Ferkol, MD will be chairing a symposium on PCD at the 2006 ATS meeting in San Diego this year. In addition to clinical and research presentations, Meghan Manion will be speaking about what it's like to grow up with PCD, providing the patient perspective for the medical professionals in attendance. To capitalize on the interest generated by Dr. Ferkol's symposium, we are planning to have a PCD exhibit booth with educational materials. ATS is attended by an estimated 15-17,000 pulmonary health professionals from all over the world, providing us with an excellent opportunity to talk about PCD!

Early in 2006, we will be holding our first conference call with the entire medical/scientific advisory board to establish future research and clinical goals. The medical/scientific advisory board is composed of physicians and researchers from disciplines related to PCD. Currently, we have representatives from adult and pediatric pulmonary medicine, ENT medicine, and ciliary genetics and ultrastructural science. We need representatives from other disciplines, as well, including neonatal and reproductive medicine or science. An updated list of medical advisors will be posted to the Foundation website at www.pcdfoundation.org or is available by contacting the Foundation. If you are interested in serving as a medical or scientific advisor, please contact the Foundation.

You may also want to mark your calendar for our Fourth Annual PCD Foundation Family Education Day to be held at the Millennium Hotel in Buffalo, NY on Saturday, August 12. Registration materials and additional information will be available soon. For more information contact Lynn Ehrne at linnie1@frontiernet.net or call the Foundation.