

THE BEAT



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Please Help With "Painless" Fundraising

Want to help, but terrified of fund- raising? Here are some painless ways to raise funds for the PCD Foundation:

1. Shop with your regular vendor and raise money at the same time! [Online shopping services that donate a percentage of sales:](#)

- *IGive: www.igive.com/
- *FreePledge: [www. Freepledge.com](http://www.Freepledge.com)

2. Search the internet like you would with any other search engine and raise funds for PCD! [Search engines that donate a penny-per-click to your designated charity:](#)

- **GoodSearch (a Yahoo affiliate): www.goodsearch.com

Clinical Genetic Test for PCD Now Available!

Clinical genetic testing for common mutations on DNAI1 and DNAH5 is now available, with physician referral, through the University of North Carolina Hospitals Molecular Genetics Laboratory. Selected exons where mutation clusters reside are sequenced. Additional mutation analysis is also available under certain circumstances. Together, mutations on DNAI1 and DNAH5 account for approximately 38% of all cases of PCD and at least 50% of PCD caused by outer dynein arm defects.

How is Genetic Testing for PCD Done?

Using a simple blood sample, DNA is analyzed for mutations known to cause PCD.

Why Consider Genetic Testing for PCD?

Genetic test results may:

- 1.) Diagnose whether or not you or your child have this condition.
- 2.) Determine whether or not family members are carriers for this condition.

When Should Genetic Testing for PCD be Considered?

- 1.) Patients with clinical disease compatible with PCD, but no defined etiology.
- 2) Patients with a history of neonatal respiratory distress—especially in term neonates.
- 3) Suppurative airways disease of unknown etiology, even with normal situs.
- 4) Persistent/chronic cough and sinusitis.
- 5) Non-CF bronchiectasis.
- 6) Severe middle ear disease.
- 7) Situs inversus totalis or situs ambiguus/heterotaxy.
- 8) Congenital heart disease with situs inversus totalis or situs ambiguus.
- 9) Non-CF male infertility in conjunction with other features of PCD.
- 10) Airways disease along with congenital heart disease, kidney disease or hydrocephalus.
- 11) A family history of PCD/KS.

What are the Limitations of Genetic Testing for PCD?

PCD is a multi-genetic disease. The current test will only identify mutations in the two genes named above, both of which code for outer dynein proteins. A negative test does not rule out mutations on other, as yet unidentified PCD-associated genes.

For more information, please contact the Molecular Genetics Lab at (919) 966-4408.

All Things PCD: A Patient's Story

by Lynn Ehrne

Have you ever wondered who the PCD Foundation is? Of course, it is all of us—patients, parents and caregivers but do you wonder who volunteers and why? In previous newsletters, you've read about Michele Manion and how the Foundation was started. Through the next few newsletters you'll hear about volunteers and what keeps the foundation going.

I thought I'd start the series off. I'm 38-years-old and was diagnosed with Kartagener's syndrome (now called PCD with situs inversus) at age nine. That was back in the 70's, long before the age of the internet and the information superhighway. How did my parents get information? They didn't. In fact they were riddled with misinformation or, more accurately, lack of information.

I was told I was born with a runny nose. It's the big joke that makes everyone laugh. How did they know I had a runny nose...aren't we covered in baby gook when we are born? Shortly after birth it was discovered that I had situs inversus totalis or more simply, organ reversal. My organs were placed as though I was looking in a mirror. Thankfully they all functioned properly. However, I'd find out years later my cilia did not. I had a constant runny nose, ear infections, upper respiratory and lung infections. My parents were told that I was just a sick kid. That was a very technical diagnosis.

My earliest memory of anything PCD-related is ear tubes. I don't remember much about the ENT, just all his scary instruments. I had mixed feelings about the ENT's office. It was the place that hurt when they took stuff out of my ears, but I could breathe and smell after I left because my nose was clear. Being able to smell fresh air was an amazing feeling every time. The things most folks take for granted were little gifts to me. I remember waking after getting my tubes in or out in the ENT's office. I was awake before my eyes were open. I could hear myself yelling and crying. My arms and legs were flailing all around. I heard the nurse say it was going to be okay and so it began...

My health problems began to escalate in grammar school. During gym class I ran and jumped off a spring board and landed hard and flat on my back. I knocked the wind out of myself. I literally couldn't breathe. I had never been so scared in my life (it even topped Friday the 13th part 3 in 3-d). I caught my breath and went home from school. It still hurt to breathe so my parents took me to the emergency room. Eventually they discovered that my left lung had collapsed. I had a localized (confined to one spot) area of bronchiectasis and the decision was made to perform a lobectomy. I was nine-years-old and very scared. When I woke up I was in intensive care and my parents were there. They asked me if I wanted anything and I remember saying "a chef salad." I think then they knew/hoped I would be okay. I had staples from the middle of my chest to the middle of my back and two chest tubes sticking out my side. It didn't hurt at first. I had around-the-clock care and when I was well enough, I was sent back to a regular room. The first night I panicked. It went from around-the-clock attention to my folks having to go to work and not being able to stay with me and less nurses with more kids to care for. I cried and told the little girl next to me I was dying. She called the nurse who very gruffly told me that I wasn't dying and that my parents had things to do. I know she meant well but I will never forget that panicked feeling.

It was a pure relief to get home. It was just before the holidays. Not sure how long after the surgery the diagnosis came but it did. My parents had no idea what Kartagener's syndrome was. All they knew was that their little girl just had a major surgery and some cruddy disease. The harder part was when they were told I would be lucky to live 6 months to a year. I overheard my mom talking on the phone, telling someone that I was dying. At nine-years-old you don't know what to do with that kind of information. I didn't tell my parents I knew. I kept it inside along with all the fear. Little bits if it would creep out in the form of tears when I was sick. I was afraid to sleep. What if I never woke up? What if I died in my sleep? What would my parents do? What would happen to my brother? It was the worst year my life. Eventually it turned out to be the best year. Lots of people come to visit you when they think you are dying AND they bring lots of gifts. The kicker to that year was that I didn't die...YAHOOO.

That's not to say that the fear ever went away. In my early teens, I found a medical article/journal about KS. It was about a 20-something-year-old who had died. All those panicked feelings rushed in again. I thought, "Wow, I survived that year and now I'm half dead again." It was hard to deal with and there was no one to talk to. I couldn't find anyone who had my disease. No one understood what it felt like. I was so angry & scared. Then my grandfather, a lifelong heavy smoker, was diagnosed with lung cancer. He went from this large as life, big guy, to a tiny, frail, pale, shell of the person he once was. He had to have a lobectomy and died shortly after from pneumonia. That freaked me out. We had the same surgery and he died. I didn't know much about cancer or most medical things back then. I related one thing to the other. I desperately hoped for someone to share all this with. At some point I headed to the library again. I found another case study of a KS patient, who died in their 40's. By this time I was married and in my very early 20's. I started to think of the half dead thing again but something made me get over myself. Maybe it was my wonderful husband and the prospect of living a good life. I decided that I wasted too much time being afraid. I was still alive after all. I mentally gave myself a kick in the pants.

Years went by and there were high and lows with my health. Thankfully the painful ear infections stopped when I was a preteen. In my 20's my lungs were persnickety as ever. Still didn't find anyone with KS. My sinuses started to get pretty bad. I was lucky with the jobs that I had, understanding bosses and all. I had to deal with the occasional co-worker that thought I was faking. If you can't see a disease, apparently it's not real. As my luck would have it, my company was sold and I lost my job but there was a silver lining to this cloud. I used my severance package to buy a computer and get online. I did a search on Kartagener's and came across a lung disease forum. I found archived posts from other KS'ers. I sat there and cried at the computer. I couldn't

Continued next page

believe my eyes. I was 31 years old and finally found evidence of others like me. I read everything I could but all the notes were old. It took me a couple days to connect with a live person. It was amazing and incredible! A few of us posted back and forth before we moved to our own group. I learned so much about my disease and the different terms that referenced it. I learned that I actually have primary ciliary dyskinesia (PCD) and that KS is the name for PCD with situs inversus. Searching for PCD, along with info from the support group led me to the UNC website. I immediately enrolled in the study. I learned about airway clearance and all these other things I never knew about. That, in turn, led me to Michele. She worked at the Vest Company but was also working hard to get the Foundation off the ground. I was so excited. After all these years there was going to be a Foundation for PCD. Finally, I had something positive to do with all the negative chronic illness energy. The first major patient event I was involved with was the 1st PCD Family Day. After a lifetime of living alone with this disease there was finally support. I was so overwhelmed. The talks from the researchers/doctors were stunning, as I never had a doctor that even knew what PCD or KS were. I met other PCD'ers. I could be myself and not worry about my "smokers-sounding" cough. I was with people who truly understood. The thing that had me in tears, though, was watching the kids. They were laughing and talking. They had each other. They didn't have to be alone. It's what motivates me. I hope no one ever feels the way I did growing up.

I've learned so much in the last several years. Participating in the research has been really eye-opening. I've participated in a couple of studies. My first visit to UNC was enlightening. I saw a slide of normal cilia and then I saw my own. It was strange to finally "see" what makes me sick. Since my first visit over 5 years ago, they have discovered the 2 genetic mutations (on DNAH5) that caused my KS. Eventually, this information will be available to most patients with PCD. Researchers are working feverishly to learn more about all potential PCD-causing mutations.

I've been in touch with many families over the years. One thing we all have in common is looking for answers. After an enormous amount of effort by PCD researchers and the PCD Foundation, we finally got the attention of the National Institutes of Health (NIH) and were funded for three basic studies. I was disheartened earlier in the summer to learn that one of the research studies is in jeopardy of being shut down because of lack of patient participation. For studies to be valid there has to be enough people participating. With such a small patient group, it is important for everyone to contribute. It is the only way we'll get the answers we are all looking for.

PCD Family Education Weekend

by Lori Ondos

This past August we made our first trip to North Carolina to attend the PCD Family Education Weekend. What a wonderful opportunity it was to finally meet Michele and her daughter and all the other families who go through the same medical issues we face on a daily basis. When Braedon, our third son, was born six years ago we were not prepared to hear that he had situs inversus and could have PCD. The doctors were honest and said they had heard of PCD but had never seen or treated anyone with it before. Getting the diagnosis was not an easy task, either. By the time we found out Braedon had PCD he had already been sick for many years. The doctors did a great job at passing the buck to each other for many years. I knew without any doubt, that despite my lack of medical background, I had to research and learn everything there was to know about this very rare genetic disorder. I felt I had to be the one to educate the doctors and suggest new things I was learning from the PCD Foundation website and from consulting with other families. That is why we attended the PCD Family Education Weekend this summer and will attend every year. I finally feel like we are not alone and Braedon was able to meet other children just like him. Attending the conference was beneficial on so many levels. The doctors from the University of North Carolina Medical Center and every person with the Foundation are so dedicated to research and to educating all of us on ways to make life livable and ways to live in this germ-filled world with compromised immune function. I feel really good about Braedon's future and about joining the PCD Foundation.



The Ondos Family at PCD Family Education Weekend, August 2007

Fundraising News

Asking for money. Just the thought of it is enough to make most of us cringe, yet the continued existence of our patient organization requires that we be willing to do just that. The Fundraising Committee of the PCD Foundation is committed to finding fund and effective fundraising activities that will engage not just the PCD community, but others outside the PCD community, as well. We recognize that our core group is small and has the additional burden of managing a chronic illness—a situation that pretty much taps financial resources. We encourage you to consider fundraising opportunities, like those listed below, to involve friends, family and neighbors in the process. We will send out periodic newsletter with ideas and experiences to motivate your creative fundraising juices!

We are looking ahead to fiscal year 2008, and need to start our fundraising efforts. Foundation funds allow us to sponsor research and educational events, provide printed and electronic materials and raise awareness of inherited ciliary disorders. Here are some fun ways you can help this Fall:

Nicholson Productions Fundraiser

Looking for unique gifts and wrap for the upcoming holiday season? Check out Nicholson Productions! From October 7 to October 21, all Nicholson Productions purchases will generate a 50% donation for the PCD Foundation. Catalogs for person-to-person sales are available from the PCD Foundation or orders can be placed online at www.nicholsonproductions.com. The code for online ordering is id1855. Please note, this code will not work until the PCD Foundation campaign starts on starting Oct 7.



PCD "A-Thons"

We have set aside the last two weeks in October and the first two weeks in November for PCD "a-thon" activities. Why not just a "walk-a-thon?" Because we want you to be able to incorporate YOUR favorite activity into an "a-thon" event. You choose the activity, walking, riding, bowling, knitting, scrapbooking, etc.—and we'll provide the support you need to have a successful event. Please contact Gina Manning for more information @ info@pcdfoundation.org.

PCD Foundation Annual Sponsor Letter

Each year we seek support from our friends and family in the form of an annual letter highlighting our activities, successes and ongoing goals. As in past years, we encourage PCD families to identify 10 - 20 friends and family members to receive this letter and suggest that participants enclose a personal note explaining how PCD has affected them and why continued research and support is so vital. This annual campaign is scheduled for the first week in November. The PCD Foundation will provide letter-head and envelopes or will mail your letters for you if you provide the addresses. If you would like to see samples from previous campaigns, please contact the PCD Foundation at info@pcdfoundation.org.

This annual campaign provides most of the operating funds the PCD Foundation needs to cover basic expenses throughout the year, plus funding research and educational initiatives. Your support of this activity is crucial and very much appreciated!

Barnes & Noble Gift-wrap Tables

Barnes and Noble stores offer free gift-wrapping during the holiday season. In return for this service, customers are asked to make a donation to a charity designated for the day or time. This fun and easy fundraiser is available to all, provided you have made arrangements with your local Barnes & Noble ahead of time. Check with your local store to see if giftwrapping slots are available in your area!

Announcement from the Fundraising Committee:

We are looking for Regional Fundraising Chairpeople to cover the following areas:

Southeast: AL, FL, GA, KY, NC, SC, TN, VA, WV
South Central: AR, LA, MS, MO, NM, OK, TX
Northwest: AK, ID, MT, OR, WA, WY

This newly created role will assist the Fundraising Committee with meeting fundraising objectives in specific regions. If you are interested, please contact Lori Truax or Heidi Burdo at info@pcdfoundation.org

Volunteers Needed From Ontario, Canada ATS Meeting May 18-20, 2008

The annual American Thoracic Society (ATS) meeting draws 16,000+ pulmonary professionals from all over the world. Next year, the ATS meeting will be held in Ontario, Canada. The PCD Foundation is going to have an educational booth at this meeting and we are looking for volunteers from the Ontario area to help us man the booth during open hours (10:00 am to 5:00 pm May 18, 19 and 20). Training will be provided.

If you are interested, please contact Michele Manion at info@pcdfoundation.org or 623-215-2032.

Good Pulmonary Toilet

By Gary Ricker

Cough and spit—that's life, ain't it? Loogie, hocker, hawk-up, lung butter, clam, pea soup, pesto—to quote the late Gilda Radner: "Did this stuff come outta me?" My lungs answer with the old snack-chip slogan: "Cough all you want, we'll make more!" It's been endless, from the Shrek-y green lines on my upper lip in grade school, to today's eight-ounce yogurt cup at my side, filled daily, usually dumped, and sometimes filled again.

My pulmonary doctor says "you're drowning in it" and tells me to find an infectious disease doctor. My Pulmonary Rehab nurse claims it's normal for smokers to cough up (and swallow—ewww!) several cups of goo every day. But I don't smoke—and swallow? Please, only on the machines in the gym, twice a week. Borrow your towel?

I'm the sputum-volume champ of my online lung chat group—I wear a green sash and tiara when I sit at my computer. Lately, that crown is heavy. Not so many years ago, after backwashing me with what seemed to be a firehose, an ear-nose-throat doc said to me, "Mr. Ricker, you're a one-man mucus factory." I've always lacked an entrepreneurial spirit; otherwise I'd have thought to ask, "Is there much call for that?"

Commercial uses, hmm—I might have started Phoenix Phlegm Farms, back when land was cheap, with low-paid workers spreading it in massive grids easily seen from the air ("Goes on sticky, dries light and flaky—Phoenix Phlegm.") Like George Washington Carver and the peanut, I'd find dozens of uses for my ever-present secretions: a slow-drying, temporary adhesive, of course; some sort of fabric coating (discovered accidentally on my shirtfront); and, as it's the right color and easily pulled into strands, an artificial turf substitute.

In an air-conditioned building, spotless in white and stainless steel (USDA approved) I'd rotate through my therapeutic regime—of course, the Vest, with its retro-looking hair-dryer hoses, and yes, a slant board for postural drainage; but what about those other, stumbled-upon ways to mobilize secretions? There'd be a TV room playing movie comedies, a comfortable recliner and a stainless-steel funnel nearby. Next door, a rolling-barrel feature as found in state-fair funhouses—collection could be a problem; I'm thinking teflon-coated Kleenex, wadded in a free hand, might serve. Since my home is currently surrounded by public-works construction (light rail and a flood retention basin), another room would have earthmoving equipment and hard-hatted men with low-slung jeans, backing up traffic for miles, rendering the air opaque. A water truck for keeping down dust would be parked nearby, doing nothing, just as in real life. Next door—what's this?—just a telephone and chair, hmm. Let's just step in here and pick it up—ah, a health insurance company's customer-service line. Always good for a few ounces—better get a blood-pressure cuff in there, too.

A hospital bed with IV tower is next door—give me a big bag of Potassium Chloride and a little breast-implant-sized pouch of antibiotic hanging up and add a pitcher of ice water to drink—the stuff flows out of me like Gatorade. Something must be done about those IV machines, though—bend an elbow and here's that little "be bawdy, be bawdy" while the digital screen flashes DOWNSTREAM OCCLUSION. I hear that alarm while watching "ER"—they use it indiscriminately, it seems, whether someone's got a line going or not—and my right arm always straightens violently. What else is at the Phlegm Farm? Oh yes, the shower: turn it on, start coughing, oh shoot, where's my cup, I don't want to clog the drain screen, it's full of hair as it is. Ugh. Nothing to do but walk wet to the toilet.

Talk about good pulmonary toilet! As long as we're in there, wouldn't a real pulmonary toilet be a great invention? Does your family scream (and flush) when confronted with an otherwise-empty commode with just a small green floater marring its placid surface? My PCD dream house (here comes Ty Pennington suppressing his gag reflex) would include several soup-bowl-sized toilets at chest height, saving gallons of water per disgusted flush.

I know it's not funny, this green river that flows from us, or worse, from our tiny, helpless loved ones, but it's there, never-ending, and if any encouragement can be drawn from a fiftyish PCD patient, it's just that—I'm still around, I can laugh about it. While I await an official diagnosis, the bittersweet thing about sitting in a room of PCDers and their parents was the prescription I'd heard so many times before. Airway clearance. Good pulmonary toilet. Mobilize secretions. Cough and spit, in other words. Well, I'm good at that, anyway. Guess I'll head out to the barn.



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Gary Ricker has written criticism and humor for newspapers in Chicago and Phoenix. Married, father of a grown daughter, his hobbies include reading, writing and pursed-lip breathing.

News About Primary Ciliary Dyskinesia

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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 large) fee, and 3.) the PCD Foundation maintains a bibliography of many articles and may be able to provide single copies for educational purposes.

- ▶ Kennedy MP, Omran H. *Congenital heart defects and other heterotaxic defects in a large cohort of patients with primary ciliary dyskinesia*. *Circulation*. 2007 June;115(22).
- ▶ Kennedy MP, Noone P, et al. *High-resolution CT scan of patients with primary ciliary dyskinesia*. *Am J Roentgenol*. 2007 May;188(5):1232-8.
- ▶ Zariwala MA, Knowles MR, Omran H. *Genetic defects in ciliary structure and function*. *Annu Rev Physiol*. 2007;69:423-50.