

THE BEAT

Summer 2008

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New Publications from the PCDF:

Gunky Lungs and Bungee Boogers: the Kid's Guide to PCD
 A 20 page, medically reviewed, illustrated booklet explaining what happens in PCD and how to treat it. Designed for ages 8 and older, but may be appropriate for younger children with parental supervision.

PCD General Brochure in Spanish

Both items available free of charge. Donations are appreciated to cover printing and postage costs.

SELL YOUR STUFF FOR PCD!!

Join us for PCDF "Garage Sale" on eBay next month. Check the website and your email for details soon.

Participation in Research: Considerations for People with PCD

A primary objective of the PCD Foundation (PCDF) is to support and eventually fund research aimed at better understanding PCD and providing optimal treatment based on that improved knowledge of the disorder. To that end, the PCDF encourages all affected individuals to consider contributing to ongoing research efforts. Right now, there are very few studies specifically devoted to PCD (see list on page 5). Three of the four studies are under the umbrella of the Genetic Disorders of Mucociliary Clearance Consortium (GDMCC), a multi-center network of seven sites in North America funded by the National Institutes of Health (NIH) through their Office of Rare Diseases (ORD). The following considerations may have general application, but they are provided mostly in reference to the GDMCC studies.

First and foremost, **participation in research is entirely voluntary**. You have no obligation to participate and there is no right or wrong decision about participation. These considerations are intended to help you make a more informed decision.

Potential Cons of Research Participation:

- You may not receive any direct personal benefit (increased knowledge, better therapeutic intervention) from participation
- There may be minimal risk involved (any potential risk will be disclosed, as required by law)
- You may incur expenses related to travel, missed work, etc. There is never a direct charge for participation in legitimate research

Potential Pros (GDMCC-specific):

- Because PCD is poorly understood and frequently misdiagnosed—even by excellent medical facilities—participation in the GDMCC studies may provide you with access to information about PCD not available from your local physicians.
- Participation may give you access to better diagnostic options for PCD, including nasal nitric oxide measurement, genetic testing and ciliary biopsy analysis from ultrastructural experts. These opportunities are very important and can provide critical information, even for people who feel comfortable with their current diagnosis.
- Participating in research is one way of establishing your genetic profile (either now, if your genetic mutation is currently known or in the future if it is not), which could be very important for identifying individuals as candidates for potential genetic therapy trials (aimed at curing the underlying defect) in the future.
- Participation may provide access to technologies not readily available from your local physician or clinic, including infant PFTs, infant HRCT and nasal NO testing.

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- Being registered for research through contact registries puts you “on the list” of individuals interested in possibly participating in future therapeutic trials aimed at actually assessing the effectiveness of various therapies to treat the symptoms of PCD. Trials of this nature will provide the information we need to finally get proven, effective treatments for the clinical problems caused by PCD.
- Finally and most importantly, individual participation is necessary for group results. We all want more information about PCD and access to better therapies and we certainly all hope for a better future for our children. The reality is that we have such a small patient group that we are just now starting to have sufficient numbers of identified patients to qualify for studies on drugs and therapies. Literally, every research participant counts. We will not reach our goals of better understanding of PCD and more effective therapies if we don't support the research efforts that will allow the necessary data to be collected. While there may be no direct benefit to any single individual, the participation of all patients is an investment in better care for the future.

Additional Things to Consider (GDMCC-specific):

1. Assessing Actual Discomfort of Participation

It is very hard to put our children through any additional discomfort and PCD kids go through a lot already. This is a legitimate concern. One way to assess the actual discomfort your child will be subjected to is to speak with parents who have participated in the trials. There are several online chat groups where this topic could be discussed or you can contact the PCDF and we can get you in touch with families willing to share their experiences.

2. Assessing Risk of Participation

Because the current GDMCC studies are primarily focused on disease characterization and natural history, there is very little inherent risk (as opposed to a drug study or other intervention where a new substance is being introduced to the body). However, there may be other risks/considerations related to some of the required testing. If you have concerns about these issues, contact the investigators directly and they can thoroughly explain the potential risk so you can make an informed decision. Contact information can be found at: <http://rarediseasesnetwork.epi.usf.edu/gdmcc/contact/index.htm>.

3. Participation is Not a “Now or Never” Thing

Maybe now is not the right time for you to participate, but you think you may be interested in the future. While most studies, including the GDMCC studies, have specific end dates, there is no “statute of limitations” on research participation in general. We certainly hope there will be many more opportunities for participation in PCD research initiatives in the future!

4. Research Participation is an Opportunity, Not a Requirement

While the PCDF supports research initiatives and feels that patients may benefit from participation, it is strictly a voluntary decision. There is no obligation to participate and deciding not to participate will in no way affect your status with the researchers or with the PCDF. Participation is simply an opportunity to contribute to future advancements in the diagnosis and treatment of people affected by PCD!

Funding for the current GDMCC studies ends in August of 2009. An application for renewal of funding for five more years has been made, however there is no guarantee it will be approved. If you are interested in participating in one of these studies, consider contacting a GDMCC research coordinator (<http://rarediseasesnetwork.epi.usf.edu/gdmcc/centers/index.htm>) in the next few months.

Concerned about travel expenses related to research participation? Free or reduced cost travel options can be arranged through Air Charity Network in collaboration with the ORD. Research participants in ORD studies are eligible. For more information, contact Marita Eddy at: meddy@mail.nih.gov or (301) 451-9646.

Education & Fun for Kids at PCDF Family Day

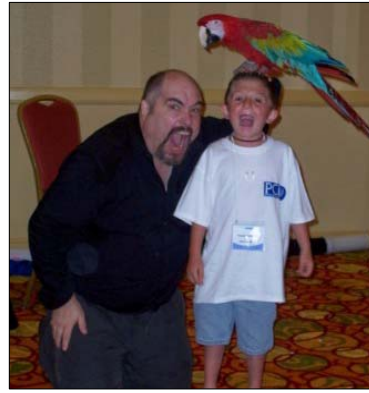
by Mary Wettengel

PCD Family Day 2008 marked the first ever "Kids' Program"! Kids who either have PCD or know someone with PCD participated.

The Saturday program began with educational presentations: *What Are Cilia Anyway?* by John Carson, PhD, UNC *Taking Care of Yourself: How to Stay Healthy* by Margaret Leigh, MD, UNC. During John Carson's session he taught the kids how normal, functioning cilia move and how PCD cilia move by separating the kids into groups and having them use their arms to create "the wave." The girls, as you can imagine, demonstrated the normal, functioning cilia and the boys demonstrated the rather "confused" cilia by waving their arms in different directions.



After some educational information, Captain Kid 'n Parrot Jack presented a magic show where all the kids got to participate and be part of the magic. Parrot Jack visited the kids at the closing of the show and even got on a few heads and shoulders! A big "thank you" goes out to the Joyner family whose donation funded Captain Kid 'n Parrot Jack.



We played our version of BINGO, called LUNGO and the kids earned PCD Bucks to win prizes. Everyone received prizes and put their prizes in their PCD "goodie" bag, along with the PCD Kids Booklet, PCD t-shirt, wristbands, and lots of other goodies. A big "thank you" to Lori Ondos for providing the PCD totes.

The Kids' Program concluded with an ice cream party that all the adults crashed! It was a sweet ending to the program and the kids reconnected with their families.

Thank you to all who helped make this program possible including Michele Manion, Lynn Ehrne, and all those mentioned already. (Editor's note: Big thank you to Paul & Mary Wettengel as well, who, along with the Joyner family, provided additional funding and assistance with organizing and managing this program)!

We're looking forward to continuing the Kids' Program and will even call this the First Annual Kids' Program!

It was just one week after my birth that I was returned to the hospital and given a diagnosis of pneumonia. This was the beginning of a life of chronic respiratory illness.

I was the boy with the runny nose and the liquid cough. They thought the wheezing and coughing were from “asthma.” Perhaps I had inherited my mother’s predisposition for sinus and lung problems, and my older sister had allergies. It was just a family thing and I would probably soon grow out of it.

I was a fairly normal child in most other ways, able to run, climb trees, swim and keep up with the other children. I grew up on a farm and there were lots of things to do like playing in the straw mound in the barn, hiking down the creek and walking through the woods. I loved to be outdoors and I especially like to climb trees. I do remember the dust in the barn causing breathing problems and I was always a mouth breather so I inhaled directly into my lungs whatever lurked in the air and coughed frequently. My nose was always stuffy and my lungs wheezy.



Dan Crow and Corky

As I grew into my teens and adulthood I began getting bouts of “bronchitis.” I would frequently cough up large amounts of mucus from my lungs and had to blow my nose with the Kleenex I always had stuffed in my pockets. The antibiotics worked well back then when there was an acute infection and I was generally only sick a few days, but the symptoms never really went away. I had sinus surgery as an adolescent, which I was told would cure my problem. It did not. I was taken to specialists in South Bend, Indiana for sinus suctioning. I would be clear on the ride home and would enjoy the open sinuses and the free breathing, but it was to be short-lived, only lasting a few hours.

I began my so-called “Cinderella” period, a time of relative good health many people with PCD experience during late adolescence and early adulthood, and was able to graduate high school and college and then begin a career as a mental health social worker.

I continued to have the same symptoms and several episodes a year of “bronchitis,” sinus infections and an occasional bout with pneumonia, but I functioned fairly well overall. I went to an allergist and took a series of shots which did nothing. Sometimes the doctors were hesitant to give me antibiotics, but they did help once I was on them. I remember a supervisor cautioning me that I was using up a lot of my sick time. I had numerous chest x-rays over the years, but the *situs inversus* and the eventual diagnosis of Kartagener’s Syndrome (PCD) went undiscovered for many years, although the doctors did note that I had scar tissue in my lungs. The disease was so rare that few doctors knew about it in the small Indiana towns I lived in.

In my frustration with the ongoing symptoms (which seemed to be getting worse) and the doctor’s inability to diagnose what was wrong, I began trying “natural cures.” I had been taking vitamins and supplements for years and reading all I could about nutrition, herbs, and natural healing. Very few things seemed to help, but I persisted. I honestly believed that I would someday find the cure for what was wrong with me and I was not going to give up until I did.

Finally, at about age 42 after living in Toledo, Ohio for several years, my family doctor sent me for a CT scan due to persistent lung infections. She called me at work and informed me that I had a rare disease named Kartagener’s Syndrome and she explained the *situs inversus* and ciliary dysfunction. I was shocked, and didn’t even yet know the full implications of this disease.

I called my partner, Bill, and told him that I had some rare disease, thinking he would probably want to get rid of me. You know, all these fears flash through your mind. I will never forget what he said to me that day, “I always knew you were one in a million.” I broke down and cried at my desk. Finally, after all these years, I knew what it was. People said, “Aren’t you glad you finally know what it is?” Not really. I mean, there was some relief, but you see, I always had this belief that I would cure myself and now I had to face up to the truth that it was not going to go away. I sometimes wonder if I didn’t stay healthier over the years simply because I did not know. All I know is that after the diagnosis, my disease began to get progressively worse.

I worked for several more years, but continued to have more serious episodes of acute lung infections and declining

pulmonary tests. I began culturing *Pseudomonas* bacteria and became resistant to many antibiotics. I was hospitalized with pneumonia in 2004 and hooked up to IV antibiotics for the first time. I thought my life was over and that maybe I should just end it. I was filled with self-pity and fear. I prayed to God and asked to be healed so many times. I began having depressive symptoms. Fortunately, I was learning all I could about the disease and got in touch with some fellow PCD folks who knew and understood what I was going through. It was so good to have their support. I met Betty and asked her how she coped with having this illness as she was older and had progressed more than I, yet seemed so determined to live as long as she could. Betty answered that being sick is all she has ever known. She was told at an early age that she would probably not live that long, yet she is now in her 60's. I asked Laura how she managed to stay so positive after the terrible periods of illness she endured and she told me how her parents were informed that she would probably not live past age 12, and that she sees every day as a gift. And there are many other wonderful people on the forum like Laurel and Lynn who have offered support. These people are survivors. They have courage and tenacity and are a gift to us all. No matter how many times they are knocked down, they pull themselves up and carry on and are there to help the others.

I made a difficult decision to go on disability in my late forties as it was just getting too hard to work with chronic lung disease. I have more time now to manage the chest percussion and postural drainage, inhalers, sinus rinses, etc. And, I continue to explore alternative healing. I am focusing on health and not so much on the disease. I exercise, eat healthy, take supplements, meditate, and am presently very involved in spiritual healing. I am working hard to halt the progression. I am still the boy with the runny nose and the liquid cough and I continue to have my pockets stuffed with tissues. On a good day I can still climb trees and I also continue to believe that there is a cure out there and that we will find it.

CURRENT PCD-SPECIFIC STUDIES SPONSORED BY THE NATIONAL INSTITUTES OF HEALTH

NIH INSTITUTE: OFFICE OF RARE DISEASES (ORD)

These three studies are collectively referred to as the Genetic Disorders of Mucociliary Clearance Consortium (GDMCC) studies and they are critically important to the patient community as they are the foundation studies required to support future PCD research efforts.

1. **Protocol 5902: Rare Genetic Disorders of the Airways: Cross-Sectional Comparison of Clinical Features and Development of Novel Screening and Genetic Tests.** Principle Investigator: Michael Knowles, MD. Designed to test large number of subjects with suspected PCD, variant CF, and PHA. Anticipate identification of enough patients with PCD through this program and clinical genetic testing to allow targeted clinical trials in future.
2. **Protocol 5901: Longitudinal Study of Primary Ciliary Dyskinesia: Participants 5-18 Years of Age.** Principle Investigator: Margaret Leigh, MD. Designed to quantitate changes in lung function, and identify associated clinical variables over 5 years.
3. **Protocol 5903: Longitudinal Study of Primary Ciliary Dyskinesia: Participants < age 5.** Co-investigators Stephanie Davis, MD and Margaret Rosenfeld, MD. Designed to define age of onset of onset changes, using pulmonary function testing and chest CTs in infants and young children over 5 years. Will be analyzed also by comparison to parallel study in CF infants/children.

NIH INSTITUTE: NATIONAL INSTITUTE OF ALLERGY & INFECTIOUS DISEASE (NIAID) (This a companion study to the three GDMCC studies listed above).

ClinicalTrials.gov Study NCT00368446: Genetic Disorders of Mucociliary Clearance in Nontuberculous Mycobacterial Lung Disease.

Principle investigator: Kenneth Olivier, MD. Healthy volunteers 18 years of age and older and patients 2 years of age or older with suspected primary ciliary dyskinesia (PCD), variant cystic fibrosis (CF) or pseudohypoaldosteronism (PHA) may be eligible for this study. Patients enrolled in the Natural History Study of Nontuberculous Mycobacteria at NIH or other NIH natural history protocols may also be enrolled. Participants undergo the following tests and procedures during a 1-day visit at the NIH Clinical Center.

NIH INSTITUTE: NATIONAL HEART, LUNG & BLOOD INSTITUTE (NHLBI)

ClinicalTrials.gov Study NCT00608556: Dyskinesia, Heterotaxy and Congenital Heart Disease. Principle investigator: Cecelia Lo, PhD. This study will examine genetic material obtained from blood and tissue samples of patients with congenital heart disease (CHD) and heterotaxy (an abnormality in the left-right positioning of organs in the body, also called situs inversus) to gain a better understanding of these disorders and of a lung disease called primary ciliary dyskinesia (PCD). CHD is prevalent in patients with heterotaxy. It is believed that certain forms of CHD or heterotaxy may have the same genetic origin as PCD.

FOR MORE INFO: <http://clinicaltrials.gov/>

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Reflections on PCDF Family Day 2008

Family

By Bill Takacs

When I think of family, my thoughts automatically turn to my biological family - my ancestors and their descendants and, of course, all of their progeny. But really, the term family has such a larger context in most of our lives. There's our work family, our church family, our extended social family - the list can go on and on.



Dan Crow, Johnny Carson and Bill Takacs at Family Day '08

This year I had the opportunity to attend the 2008 Family Day program in Durham, North Carolina, I discovered that there is yet another family dimension in my life - the PCD family!

I am the partner of Dan Crow who was diagnosed with PCD about five years ago as an adult. Dan and I have been together for fourteen years and prior to his diagnosis, he had told me about his history of having illnesses like bronchitis and even pneumonia since childhood. As the years have passed, I had observed him struggling with his "condition" - the constant coughing and clearing, the frequent fevers, and bouts of sickness that for some reason just seemed foreign to me. And then he was diagnosed. I remember him telling me that he had situs inversus with his PCD and to this day recall my utter amazement that no one had properly diagnosed that aspect of his condition until he was well into his 40's.

Prior to attending the Family Day program this year, I had acquired a rather superficial understanding of the disease largely based on information that Dan had learned and had passed on to me. Terms like "cilia," "situs inversus," "postural drainage," "pseudomonas," "IV antibiotics" and "picc lines" had all become familiar to me since Dan was diagnosed although, honestly, I really didn't comprehend the full nature and extent of what PCD is and what PCD sufferers endure until I attended Family Day this year. I knew it was serious and I thought it was a disease akin to cystic fibrosis - and that was about the full extent of my knowledge.

Having participated in Family Day, I now feel that my knowledge base is much broader. I had the chance to learn so much information about genetic predispositions, diagnostic tests and evaluations, daily maintenance programs, updates on treatments, and, of course, the importance of being a supportive partner as Dan faces his challenges so courageously on a daily basis.

Aside from the acquired knowledge, though, the Family Day program had another equally meaningful reward - family! I had the opportunity to meet others who deal with PCD on a daily basis - as patients, parents, spouses, and treatment professionals.

As I reflect back on that weekend in July, I am encouraged by the level of commitment, competence, and compassion from the doctors and staff at the University of North Carolina. I am humbled by the courage of Jack and his wife following his lung transplant. I am impressed by the activism of the family from Pennsylvania in their fundraising efforts. I am impressed by the efforts of Michelle and Lynn and the other directors whom I had the privilege of meeting. I grin at the memory of the children and all of that energy. And, most of all, I am so proud of Dan for the courage that he continues to display each and every day in dealing with his disease and in educating and supporting others.

As heinous as the disease of PCD is, I left that weekend in July uplifted, encouraged, and somehow reinforced with the knowledge and understanding that Dan and I are not alone in this ordeal. PCD is the common thread that brought all of us together for that brief weekend in July, 2008, and it is and will continue to be what makes all of us a part of the PCD family.

Family Day 2008

Family Day 2008 was a wonderful experience with many new families participating this year. For those who were unable to attend, here is the agenda a photo of the group. Many thanks to all the speakers and to the PCD families who helped make this another successful event!

8:50 – 9:00	PCD Foundation Welcome	Michele Manion
9:00 – 9:45	What is PCD?: Recent Research Progress and Prospects for the Future.	Mike Knowles, MD
9:45 -- 10:30	Ciliary Ultrastructure & Function: Assessing Ciliary Defects in PCD	John Carson, PhD
10:45 – 11:30	Adults with PCD: Advanced Lung Disease & Transplant: Current “Best Practices”	Peadar Noone, MD
11:30—12:15	Preventing Airway Damage in PCD: Approaches to Therapy	Margaret Leigh, MD
1:15 – 2:00	PCD Genes: Identifying Candidates & Developing Gene Tests	Maimoona Zariwala, PhD
2:00--2:45	Nasal Nitric Oxide (NO) and PCD	Milan Hazucha, MD
2:45—3:30	ENT Concerns in PCD	Margaret Leigh, MD
3:45—4:30	Airway Clearance Options	Jane Hess, RT
4:30—5:30	Open Question Time- Patient discussion with physician moderators	
Sunday 8:00—Noon	Group Discussion	



Resource Pages

The materials provided on this page and the following page are PCDF resources for use with your physician and with your child's teacher. Downloadable versions of both of these items are available on the website at www.pcdfoundation.org.

[Date]

Dear [insert teacher's name],

My child, [insert child's name], will be a student in your classroom this year. [Child's name] has a rare genetic disorder called primary ciliary dyskinesia (PCD). PCD is a multi-system disorder, but chronic infections of the upper and lower respiratory tract, including the lungs, the ears and the sinuses, are the primary problems [child's name] will encounter.

Cilia are microscopic projections from the tissue lining certain areas of the body, including the lungs, sinuses, ears, ventricles of the brain and the reproductive organs. While cilia may look like tiny hairs, they are actually very complex structures that perform important roles in the body. In some areas of the body, the sweeping movement of the cilia is crucial to the proper cleaning and functioning of organ systems.

In PCD the sweeping motion of the cilia is impaired because of an inherited defect. When this happens, areas that rely on ciliary activity for proper function, like the lungs, ears and sinuses, are subject to repeated infections.

You may notice that [child's name] coughs a lot. Sometimes his/her cough can sound alarming, however it is rarely contagious. Without functioning cilia, mucus that would normally be "swept" up the respiratory tract to be swallowed collects in the airways. Trapped mucus is a source for repeated infections. For people with PCD, a healthy productive cough is essential for moving mucus out of the airways. We encourage [child's name] to cough and will provide tissues for him/her to use.

People with PCD are susceptible to opportunistic infection and may get sick from bugs that don't usually infect healthy people. This means that they are generally more at risk to catch something from others than to pass something on. We stress the importance of hand washing with [child's name] and discourage him/her from close contact with sick individuals. A cold for a healthy child can easily become pneumonia for a child with PCD, so reasonable precautions to prevent the spread of contagious illnesses is very much appreciated.

Hearing loss is a common consequence of chronic ear infections in PCD. We monitor [child's name] hearing closely, but at times fluid can collect behind the ear drum and cause subtle hearing changes that he/she may not be aware of. If you notice changes consistent with hearing problems like lip-reading, talking loudly, failing to respond when called, etc., please let us know. If possible, a seating assignment close to where the teacher presents information would be helpful.

PCD frequently causes severe sinus disease. Sinus pain and infection are common consequences of the disorder. When [child's name] has active sinus disease, he/she will need to blow his/her nose frequently. Because of the nature and volume of the mucus PCD people produce, this is often an embarrassing procedure for PCD kids. We request that [child's name] be excused from the classroom when it is necessary for him/her to blow his/her nose.

Because of repeated infections, kids with PCD may have multiple absences from school. We will work with you to try to minimize the number of absences and to help with any make up work that may be required due to absences.

Roughly 50% of people with PCD have issues with organ placement (*situs*). Most have a condition called *situs inversus totalis* where all of the abdominal and chest organs are reversed. When this happens the disorder is sometimes called "Kartagener syndrome." Less frequently, people with PCD may have random issues with specific organs. [Child's name] has [fill in specifics if needed]. Usually, there are no significant problems with organ function in PCD, but we would be happy to discuss [child's name] specific concerns with you.



PCD Action Plan

Adapted from UNC, Chapel Hill PCD Action Plan



OPTIMIZING YOUR LUNG HEALTH

Your FEV1 today = _____ % predicted

Contact Information

For **clinical** questions call _____ and ask to speak to your doctor's nurse.
 For **Research** info call Susan Minnix at 919-843-3508.

AIRWAY CLEARANCE

This is the most important thing that you can do to keep your lungs healthy.

You should do airway clearance at least _____ times each day. If you have more cough than usual, increase this to 3 to 4 times each day. Avoid cough suppressants. In PCD cough is the primary form of airway clearance. Participate in vigorous aerobic activity. Avoid exposure to tobacco smoke and irritants that may increase mucus secretions. We encourage an active lifestyle. Exercise helps to loosen secretions.

STOPPING THE SPREAD OF GERMS

- Avoid contact with sick people.
- Wash your hands often.
- Get a flu shot in the fall of every year.
- Get a pneumococcal vaccine every 5 years.
- Make sure your immunizations are up to date.
- Disinfect your nebulizer equipment as instructed.

- Respiratory hygiene reviewed
- Nebulizer cup cleaning techniques reviewed

KNOW YOUR ORGANISMS

You are currently on isolation: none contact
 Your last sputum culture grew: _____

Infection control guidelines reviewed

YOUR NEBULIZED MEDICATIONS SHOULD BE IN THE FOLLOWING ORDER:

1. _____
2. _____

Your personalized plan includes the following (circle):

HUFFING VEST** ACAPELLA FLUTTER
 BUBBLES PEP CHEST PT EXERCISE

Exercise must equal 20minutes of vigorous activity.

**Vest Settings: _____ frequency, _____ pressure
 and _____ minutes per treatment.

Post airway clearance nebs:

MEDICATIONS

- Use separate nebulizer cups for each medication.
- For TOBI, only use the Pari LC Plus neb cup.
- Always take inhaled Tobi AFTER you have taken your albuterol and finished Chest PT.
- When taking Cipro or Septra avoid the sun. You may burn more easily.

Current medications:

1. _____
2. _____
3. _____
4. _____

Changes to your medications today include:

KNOW YOUR CILIARY DEFECT

- _____ Outer dynein arm
- _____ Inner dynein arm
- _____ Central apparatus defect

Notes:

Braedon Ondos Fundraiser for the PCDF – A Huge Success!

Mary Wettengel & Lori Ondos

Lori Ondos, Braedon's mom and his most dedicated advocate, coordinated a fundraising event on Saturday, August 16th at the Giant Eagle grocery store parking lot in McMurray, PA. "Quinn & Rose," popular Pittsburgh radio celebrities, did a live broadcast from the fundraising site, continually providing event information throughout the day. Rose Tennet interviewed Lori about Braedon's experiences, PCD and the event. Rose is a huge advocate for cystic fibrosis and raises a lot of money for the CF Foundation, so we are very lucky to have her on our side.



Lori Ondos and Rose Tennet broadcast on "Quinn & Rose"

Channel 4 (WTAC) Action News interviewed Lori, giving her the opportunity to spread awareness about PCD on the local news.

The outpouring from the Peters Township community was overwhelming! What started as a small fundraiser grew into an all out event. Lori's original goal was to raise \$2,500 and PCD awareness; however, very quickly Lori's goal changed to \$10,000 and after publishing two articles about the event in the newspaper and two in local magazines, the Ondos mailbox was flooded with checks and auctions items. Before it even started they had reached their \$10,000 goal!



Lori & Braedon Ondos

Volunteers from the community contributed time and services, in addition to cash and auction donations. Strive Fit Gym donated a brand new \$8,000 treadmill, along with 2 one-year memberships worth \$2,000 each. An area dentist donated an \$800 Zoom White treatment. The Pittsburgh Steelers provided baskets with tickets and t-shirts, golf packages from country clubs, and a representative of their organization came the day of the event and donated an additional 6 Pittsburgh Steelers' tickets. In all, over 70 items were donated for auction by local merchants including gift certificates to local restaurants, jewelry, handbags, Webkinz, an outdoor grill, and many more! Giant Eagle grocery store donated all the hot dogs, buns, and condiments, along with many cases of water to sell throughout the event and raise funds and Italian Village pizzeria donated half of their pizza sales.



Lori speaks with Channel 4 Action News



Volunteer Patrick Wettengel, models a balloon ensemble!

Several members of the Washington Wild Things baseball team, including their manager and mascot, supported the event by signing autographs, talking with fans, and selling 50/50 raffle tickets. Fun activities like a Euro Bungee jump, a bounce house, amazing balloons from "Mike the Balloon Guy," pony rides, fireman robot and truck tours from the local fire department, face painting, portrait painting and carnival-like games were available all day. The Spa-Nique bus offered massages and sugar scrubs in mobile luxury. Entertainment was also a big part of the day with performances by "The Number Men," with local talent Gretta Shepardson on vocals, and the Arthur Murray dance studio dancers.

The day closed with a "Prayers to Heaven" ceremony where everyone released into the sky white balloons with prayers for Braedon. There wasn't a dry eye!

Everyone can do their part in raising PCD awareness and help fund a cure for this poorly understood disease. Just ask Lori ... she never imagined the fundraiser would turn into such an amazing, successful, emotional, supportive event and now more than \$20,000 has been raised to help fund the cure! **Please help in raising PCD awareness and "funding the cure!"**

Continued from page 5.

What is Research?

“Research” is a broad term that encompasses many different aspects of scientific discovery. In medicine, research is a systematic way to evaluate traits or effects that are observed under controlled conditions. The goal of this controlled observation and analysis is to reach validated conclusions about the nature of a disorder, the safety or effectiveness of a particular intervention, the best course of treatment or prevention, or to collect data that will allow investigators to formulate new research objectives.

The Challenge for Rare Disorders

The notion of validation is critical. It is not enough that “research” be done. It must be done using standards agreed up on by the scientific community in order to achieve broad acceptance. Without the acceptance of the scientific community, it is very unlikely that a treatment or prevention would become standard of care or that insurance providers would cover it. Generally, achieving validity requires the involvement of large numbers of clearly identified individuals to demonstrate that the conclusions reached truly can be applied to the group being studied. This requires 1.) Accurate identification of affected individuals to participate in a study (i.e. a PCD study that includes people who have been misdiagnosed with PCD, but who actually have secondary ciliary issues would not produce valid information about PCD), and 2.) Enough participants to produce results that are statistically valid and that can be replicated. These requirements are particularly difficult for rare and difficult-to-diagnose disorders to meet.

Clinical versus Basic Science Research

In broad terms, clinical research refers to research done on human subjects with the goal of understanding the natural history of a disease, validating diagnostic tools, or evaluating a treatment or prevention. Basic science research generally refers to studies that attempt to answer more fundamental, but equally important, questions that may be used as the foundation for further research. For affected individuals, the importance of clinical research is obvious, but it would be a mistake to discount the importance of basic science research. In PCD for example, the genetic mutations that cause the human disease were discovered in large part by basic science research done on a single celled organism called *Chlamydomonas*. These discoveries were then *translated* (“translational research” is another term you may have heard) for use in human genetic testing. Ideally, targeted basic science research and clinical research are both employed in understanding a disease.

Creating a Scientific Foundation for Further Research

We would all be skeptical if a friend with no medical background offered to do brain surgery on us, right? The reason for this is that we understand that there is a significant foundation of knowledge and skill that must be achieved for mastery of something as complex as brain surgery. Learning about cells and biology allows a student to move on to basic anatomy and physiology, which in turn allows more in-depth study into brain anatomy and physiology, and so forth. We would not have much faith in a practitioner who skipped any of these crucial steps. The same is true in research. For the results to be valid, a solid foundation of fundamental knowledge needs to be built first. In PCD, we are well into the process of building that foundation through NIH-sponsored GDMCC studies (see page 5). Protocol 5902 attempts to clearly define what PCD is and how it can be accurately diagnosed. This is not as simple as it sounds and PCD continues to be significantly misdiagnosed and misunderstood. Protocol 5902 attempts to address those issues, as well as identify a large enough confirmed patient base for future clinical trials. Protocols 5901 and 5903 are aimed at providing clear evidence of the natural history and progression of disease in PCD. We must have this evidence of what can be expected to happen in PCD for trials of therapies or preventive measures to have any meaning. As an example, if we want to do a trial for therapeutic prevention of the acquisition of *Pseudomonas* in PCD patients, first we have to have documented evidence that *Pseudomonas* is actually acquired by a significant proportion of people with PCD. Building this foundation is expensive and time-consuming, but it is essential. The support of the PCD community in facilitating these trials is critical to advancing future research efforts.

PCD Foundation Board Election September 2008. The election for new PCDF board members was held on Monday, September 15, 2008.

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