

Overview: The conference entitled *Primary Ciliary Dyskinesia and Overlapping Syndromes* was held at the Eric P. Newman Education Center (EPNEC), which is located adjacent to the Washington University Medical Center in St. Louis, Missouri. This 2 day conference occurred on September 30th and October 1st, 2010. Approximately 70 persons attended the conference. The objectives of this conference were (1) To optimize diagnosis of PCD through standardization of diagnostic testing; (2) To define PCD genes and gene mutations through global networking; (3) To optimize clinical care of PCD patients and develop clinical research networks to test therapies through clinical trials and (4) To refine nomenclature for ciliopathies and better define overlapping features. The agenda for the conference is below. Fourteen posters were presented at the Poster Session.

DAY 1 (9/30/10)

Welcome and Introduction: 8:00-8:30 AM

Michael Knowles, MD - University of North Carolina at Chapel Hill, Department of Medicine
Stephanie Davis, MD-University of North Carolina at Chapel Hill, Department of Pediatrics

Early Morning Session: Cilia and models for studying structure and function

Section Chair: Michael Knowles, MD

8:30-9:00 - Structure and Proteomic Analysis of the Human Cilium

Larry Ostrowski, PhD - University of North Carolina at Chapel Hill, Cell/ Developmental Biology

9:05- 9:35 - Lessons from Chlamydomonas

Susan Dutcher, PhD - Washington University School of Medicine, Department of Genetics

9:40- 10:10 - Lessons from the DNAH5 Mouse Model

Cecilia Lo, PhD - University of Pittsburgh, Department of Developmental Biology

Break: 10:10-10:30 AM

Late Morning Session: The Challenges of Diagnosing PCD

Section Chair: Stephanie Davis, MD

10:30-11:00 - Ciliary Ultrastructure: Gold standard for diagnosis?

Michael Knowles, MD - University of North Carolina at Chapel Hill, Department of Medicine

11:05 – 11:35 - Role of Ciliary Beat

Christopher O'Callaghan, MD- University of Leicester, Department of Infection, Immunity and Inflammation

11:40 – 12:10 - Nasal Nitric Oxide: Utility as a screening tool and/or adjunctive test?

Margaret Leigh, MD - University of North Carolina at Chapel Hill, Department of Pediatrics

Early Afternoon session: The Genetics of PCD

Section Chair: Margaret Leigh, MD

1:30 – 2:25 - The emerging genetics of primary ciliary dyskinesia

Heymut Omran, MD – University Hospital Muenster, Department of Pediatrics

Maimoona Zariwala, PhD- University of North Carolina at Chapel Hill, Pathology and Laboratory Medicine

2:30 – 3:00 - PCD genes: Lessons from the Amish populations

Thomas Ferkol, MD - Washington University School of Medicine, Department of Pediatrics

Late Afternoon Session: Update of respiratory tract disease in PCD

Section Chair: Thomas Ferkol, MD

3:15- 3:45 - Overview of PCD-related lung disease: What happens when lung defenses fail?

Scott Sagel, MD - University of Colorado, Department of Pediatrics

3:50 – 4:20 - Early lung disease in young children with PCD

Stephanie Davis, MD - University of North Carolina at Chapel Hill, Department of Pediatrics

4:25 – 4:55 - Otolaryngologic manifestations of PCD

Paolo Campisi, MD - Toronto SickKids, Department of Otolaryngology

5:00 – 5:30 - The clinical approach to lung disease in PCD: Is there evidence?

Sharon Dell, MD - Toronto SickKids, Department of Pediatrics

Dinner: 6:00-8:00 PM - Living with PCD: A Perspective from the PCD Foundation

Michele Manion (Parent of child with PCD)

Serena Ramos (Parent of child with PCD)

Meghan Blair

Day 2 (10/01/10)

Early Morning session: Other ciliopathies with some clinical features of PCD

Section Chair: Ken Olivier, MD

8:00-8:30 - Spectrum of Clinical Diseases Caused by Other Ciliopathies

Meral Gunay-Aygun, MD, National Human Genome Research Institute

8:35 – 9:05 - Cystic kidney disease and nephronophthisis-related ciliopathies: Gene identification by exome capture and NextGen sequencing

Friedhelm Hildebrandt, MD - Howard Hughes Medical Institute, University of Michigan

9:10 – 9:40 - The Genetics of Heterotaxy Spectrum Disorders

Stephanie Ware, MD, PhD - Cincinnati Children's Hospital Medical Center.

Late Morning Session: Breakout sessions for future research/collaboration

10:00 – 12:00

Optimizing diagnosis of PCD through standardization of diagnostic testing

Chair(s): Mike Knowles, MD and Margaret Leigh, MD

Defining PCD genes and gene mutations through global networking

Chair (s): Maimoona Zariwala, PhD and Heymut Omran, MD

Optimizing clinical care of PCD patients and developing clinical research network to test therapies through clinical trials

Chair (s): Scott Sagel, MD, Stephanie Davis, MD

Refining nomenclature for ciliopathies and defining overlapping features

Chair (s): Meral Gunay-Aygun, MD and Friedhelm Hildebrandt, MD

12:00-2:00 PM Lunch and Poster Session

Reports from each of the breakout sessions:

2:00 – 2:30 - Refining Nomenclature for Ciliopathies

2:30-3:00 - Optimizing diagnosis of PCD through standardization of diagnostic testing

3:15 – 3:45 – Defining PCD genes and gene mutations through global networking

3:45 – 4:15 - Optimizing clinical care of PCD patients and developing clinical research network to test therapies through clinical trials

4:15 Closing Remarks Michael Knowles, MD

Evaluations: After the conference, evaluations were sent to the attendees. Overall, the evaluation grades were high and revealed that the conference was very well received. The evaluation grading was on a 1 to 5 scale. The responses were scored as noted below:

- 5- Strongly Agree
- 4- Agree
- 3- Neutral
- 2- Disagree
- 1- Strongly Disagree

See evaluation responses below:

Science/Ideas:

1. Presentations defined and analyzed the most important ideas & opportunities. **4.4**
2. Presentations included a substantial amount of unpublished work. **4.2**
3. Presentations were at the frontier of the field. **4.5**
4. The conference was thought provoking, stimulating, exciting. **4.6**
5. Poster session contributed unpublished research at the frontier of the field. **4.3**

Discussion at breakouts and posters:

1. Adequate time for the discussion was allowed. **4.1**
2. One person or group did not overly dominate discussion. **4.2**
3. Discussion Leader managed their session well and stimulated discussion. **4.5**
4. Discussions evoked and explored new directions. **4.6**

Management/Organization:

1. Good selection of topics **4.7**
2. Good selection of speakers **4.8**
3. Speakers were present and available for discussion after their presentation. **4.4**
4. Attendees were diverse and reflected the composition of this field. **4.3**
5. Pool of Speakers was diverse and reflected the composition of this field. **4.6**

Atmosphere:

1. The conference was more than just a meeting, workshop or collection of papers. **4.8**
2. Opportunities were available for networking with colleagues. **4.7**
3. The overall conference atmosphere was friendly, not cliquish. **4.9**
4. Informal interactions contributed strongly to the quality of the meeting. **4.5**

Overall Conference Suitability:

1. The conference met my expectations. **4.8**
2. I expect to attend this conference again. **4.7**
3. This was the best conference in the field I attended this year. **4.7**

Final Product: The speakers are currently putting together summaries for eventual publication in the *Proceedings of the American Thoracic Society*. The editor, Dr. Alan Leff, has approved publication of these summaries in this journal and the publications should go into print by the spring of 2011. Guest editors for the series will be Stephanie Davis, Margaret Leigh and Michael Knowles.

The articles will be the following:

- a) *Cilia and models for studying structure and function:*
Larry Ostrowski, Susan Dutcher and Cecilia Lo
- b) *The Challenges of Diagnosing PCD:*
Michael Knowles, Christopher O'Callaghan, Margaret Leigh
- c) *The Genetics of PCD:*
Heymut Omran, Maimoona Zariwala, Thomas Ferkol
- d) *Update of respiratory tract disease in PCD:*
Scott Sagel, Stephanie Davis, Paolo Campisi, Sharon Dell
- e) *Other ciliopathies with some clinical features of PCD:*
Meral Gunay-Aygun, Friedhelm Hildebrandt, Stephanie Ware

Collaborations established secondary to conference: Several collaborations were developed due to the conference. These collaborations include: 1) The UNC consortium is working with Dr. Friedhelm Hildebrandt to whole exome sequence related pts with PCD, or similar phenotype; 2) There is an exchange of data among Dr. Susan Dutcher's lab, Dr. Cecilia Lo's lab and the UNC group in the areas of cilia, heterotaxy, and genetic defects in the Amish as well as those afflicted with congenital heart disease; 3) The Mucociliary Clearance Consortium is working with Dr. Omran to further evaluate two new PCD genes from Europe; 4) Dr. Zariwala and Dr. Omran are working on a novel gene recently discovered at UNC; 5) Dr. Friedhelm Hildebrandt and Dr. Zariwala are evaluating PCD pts for mutations found in patients with Joubert's for possible "crossover." and 6) An international clinical trials network may also be developed amongst the group of investigators at the meeting.