Conference: Primary Ciliary Dyskinesia and Overlapping Syndromes

DAY 1:

Welcome and Introduction
Mike 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: 8:15-10:30 AM - Cilia and models for studying structure and function

Structure and Proteomic Analysis of the Human Cilium
Larry Ostrowski, PhD - University of North Carolina at Chapel Hill, Cell/ Developmental Biology

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

Lessons from the DNAH5 Mouse Model
Cecilia Lo, PhD - University of Pittsburgh, Department of Developmental Biolog
Break: 10:30-10:45 AM

Late Morning Session 10:45-12:15 - The Challenges of Diagnosing PCD

Ciliary Ultrastructure: Gold standard for diagnosis?
Mike Knowles, MD - University of North Carolina at Chapel Hill, Department of Medicine

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

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

Lunch: 12:15-1:30PM

Early Afternoon session: 1:30-3:00 PM - The Genetics of PCD

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

PCD genes: Lessons from the Amish populations
Thomas Ferkol, MD - Washington University School of Medicine, Department of Pediatrics

3:00-3:15PM Break

Late Afternoon Session: 3:00-5:15 PM - Update of respiratory tract disease in PCD

Overview of PCD-related lung disease: What happens when lung defenses fail?
Scott Sagel, MD - University of Colorado, Department of Pediatrics

Early lung disease in young children with PCD
Stephanie Davis, MD - University of North Carolina at Chapel Hill, Department of Pediatrics

Otolaryngologic manifestations of PCD
Paolo Campisi, MD - Toronto SickKids, Department of Otolaryngology

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 with child with PCD)
Carey Kaufmann (Parent with child with PCD)
Lynn Ehrne (Adult with PCD)

Day 2:

Early Morning session 8:00-9:30 AM - Other ciliopathies with some clinical features of PCD

Spectrum of Clinical Diseases Caused by Other Cilopathies
Meral Gunay-Aygun, MD, National Human Genome Research Institute

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

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

9:30-10:00 AM Break

Late Morning Session 10:00-12:00AM - Breakout sessions for future research/collaboration

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

12:00-1:30 PM Lunch and Poster Session

1:30-4:30 Reports from each of the breakout sessions

4:30 Closing Remarks Mike Knowles, MD