






2019 USCAP Companion Meeting

*Sunday, March 17, 2019 – 8:30am-12:00 noon
Gaylord National Resort and Convention Center
National Harbor, Maryland, USA*

Primary Ciliopathies: Diagnosing Motile and Non-motile Ciliary Disorders

Genetic disorders affecting the structure and/or function of cilia belong to an emerging class of disease processes termed ciliopathies. Primary ciliary dyskinesia refers to a heterogeneous collection of disease-causing mutations which result in impaired cilia function leading to a constellation of symptoms including chronic airway disease, laterality anomalies, and fertility issues. The diagnosis of PCD has historically relied on the assessment of clinical features combined with the identification of hallmark ultrastructural abnormalities; however, the evaluation of cilia biopsies by TEM presents both technical and interpretive challenges. The recent availability of commercial genetic testing panels as well as the development of several new screening tests has greatly improved the ability to make a timely and accurate diagnosis. As no single test is 100% effective in diagnosing PCD, this session will focus on the ultrastructural features, the emerging technologies available for diagnosing PCD, and the international consensus of PCD diagnostic guidelines. A review of the pathophysiology and diagnosis of polycystic renal disease and research on cilia-exosome interaction will be presented.

PRESENTATIONS

Times	Title	Presenters
8:30 am – 9:30 am	PCD: The Correlation of EM Features with New and Established PCD Diagnostic Techniques	Kyriacos Kyriacou, PhD, FRMSoc The Cyprus Institute of Neurology and Genetics Nicosia, Cyprus 
9:30 am – 10:00 am	An Australian Experience of PCD Diagnosis Including the Use of Immunofluorescence as a Useful Adjunct to Transmission Electron Microscopy	Louise Hughes, MBBS FRCPA Concord Repatriation General Hospital Sydney, Australia 
10:00 am – 10:30 am	Coffee Break	Juan Valdez, et al ¡Disfrute de un buen café! 

10:30 am – 11:15 am Pathology and Cyst Origin in PKD

**Timothy Fields,
MD, PhD**

University of
Kansas Medical
Center
Kansas City, KS,
USA



11:15 am – 12:00 noon The Cilia-Exosome Interaction

**Christopher
Ward, MB,
ChB, PhD**

University of
Kansas Medical
Center
Kansas City, KS,
USA



Moderators

Giovanna M. Crisi, MD, PhD

Baystate Health
Springfield, MA, USA



Eric P. Wartchow, PhD(c)

Children's Hospital Colorado
Aurora, CO, USA

