



**DEPARTMENT  
OF PEDIATRICS**



**THE ISRAELI SOCIETY FOR PEDIATRIC PULMONOLOGY**

**INTERNATIONAL RESEARCH CONFERENCE ON**

**CLINICAL AND MOLECULAR ASPECTS  
OF PRIMARY CILIARY DYSKINESIA (PCD)**

**29-30 January 2014**

**All lectures will take place at  
the Feldman Building, on the Edmond J. Safra, Givat Ram Campus**

**Organizing committee:**

**Eitan Kerem, MD**, Hadassah-Hebrew University Medical Center, Jerusalem

**Thomas Ferkol, MD**, Washington University in St Louis, St. Louis, USA

**Israel Amirav MD**, Ziv Medical Center, Israel

**Steven Brody, MD**, Washington University in St Louis, St. Louis, USA

## PROGRAM

### Wednesday, 29 January 2014

08:30 – 09:00 Registration and Coffee

09:00 – 09:15 *Greetings*

**Eitan Kerem MD**, Hadassah Hebrew University Medical Center, Israel

**Michal Linial**, IIAS Director, Israel

**Thomas Ferkol MD**, Washington University School of Medicine, USA

**Session 1** *Clinical Features of Primary Ciliary Dyskinesia and other Ciliopathies*

**Session Chairs: Prof. Asher Tal, MD & Prof. Neville Berkman, MD**

09:15 – 09:40 **Thomas Ferkol, MD**, Washington University School of Medicine, USA,  
*Clinical Concepts in Ciliopathies*

09:40 – 09:50 Discussion

09:50 – 10:15 **Angelo Barbato MD**, University of Padua, Italy  
*Treatment Approach to PCD in Children*

10:15 – 10:25 Discussion

10:15 – 10:35 **Israel Amirav MD**, Ziv Medical Center, Israel, *The Israeli Consortium for PCD*

10:35 – 10:45 Discussion

10:45 – 11:00 Coffee Break

11:00 – 11:20 **Simon Edvardson MD**, Hadassah Hebrew University Medical Center,  
Israel, *Spectrum of Clinical Diseases Caused by Primary Ciliopathies*

11:20 – 11:30 Discussion

11:30 – 11:45 **Malena Cohen Cymberknoh, MD**, Hadassah Hebrew University Medical  
Center, Israel, *Clinical Features of Primary Ciliary Dyskinesia: Comparison with Cystic  
Fibrosis*

11:45 – 11:55 Discussion

12:00 – 13:00 Lunch Break

**Session 2** *Cilia Genes, Proteins and Ciliogenesis*

**Session Chairs: Amir Kugelman, MD & David Shoseyov, MD**

13:00-13:20 **Heymut Omran, MD**, University Hospital Muenster, Germany  
*Structure and Proteomic Analysis of the Human Cilium*

13:20-13:30 Discussion

13:30-13:50 **Steven Brody, MD** Washington University School of Medicine, USA  
*Modeling Regulation of Ciliogenesis in Primary Airway Epithelial Cells*

13:50-14:00 Discussion

14:00 – 14:10 Coffee Break

14:10 -14:30 **Susan Dutcher, PhD**, Washington University School of Medicine, USA  
*Lessons Learned from Green Alga - Chlamydomonas Reinhardtii*

14:30-14:40 Discussion

14:40-15:00 **Amjad Horani, MD**, Washington University School of Medicine, USA  
*Use of Primary Human Airway Epithelial Cells to Model PCD Phenotypes*

15:00-15:10 Discussion

15:10-15:25 **David Shoseyov MD**, Hadassah Hebrew University Medical Center, Israel,  
*Cilia Beat Frequency Measurements and Waveform Analysis – Pitfalls and Advances*

15:25-15:35 Discussion

15:35 Tour of Jerusalem

## **Thursday 30 January 2014**

08:30 – 09:00 Coffee

### **Session 3** *Diagnostic Criteria and Genetic Testing for PCD*

#### **Session Chairs: Orly Elpeleg, MD & Amihud Singer, MD**

09:00 – 09:20 **Huda Mussafi MD**, Schneider's Children Medical Center, Petah Tikvah  
*The European Task Force Project and the Israeli Experience*

09:20 – 09:30 Discussion

9:30-9:50 **Kim Nielsen, MD**, Copenhagen University Hospital, Denmark  
*Nasal NO in PCD – What is it Good For?*

9:50 – 10:00 Discussion

10:00 – 10:20 **Orly Elpeleg MD**, Hadassah Hebrew University Medical Center, Israel  
*The Exome Revolution in Diagnosing Rare Inherited Pediatric Disease*

10:20 – 10:30 Discussion

10:30 – 10:50 **Heymut Omran, MD**, University Hospital Muenster, Germany  
*Genetic Testing in Primary Ciliary Dyskinesia*

10:50 – 11:00 Discussion

11:00 - 11:15 Coffee Break

11:15 – 11:35 **Hannah Mitchison, PhD**, University College London (UCL), UK,  
*Genetic Analysis of PCD, Druze Population and Beyond*

11:35 – 11:45 Discussion

11:45 – 12:05 **Eitan Kerem MD**, Hadassah Hebrew University Medical Center, Israel  
*New Diagnostic Criteria for Primary Ciliary Dyskinesia? The Experience from the Hadassah PCD Clinic*

12:05 – 12:15 Discussion

12:15 – 13:00 Lunch Break

**Session 4** *PCD, Laterality Defects and Novel Gene Candidates*

**Session Chairs: Amjad Horani, MD & Amnon Ariel, MD**

(20 minute Talks, 10 minute Discussion)

13:00 – 13:20 **Steven Brody, MD**, Washington University School of Medicine, Department of Medicine, *Animal Models of PCD and Laterality Defects*

13:20-13:30 Discussion

13:30 – 13:50 **Yuval Cinnamon PhD**, Hadassah Hebrew University Medical Center, Israel, *Mechanism of Lateralization*

13:50-14:00 Discussion

14:00 – 14:10 Coffee Break

14:10 -14:30 **Ruth Parvari PhD**, Ben-Gurion University of the Negev, Israel, *Search for Genes Mutated in Lateralization Defects*

14:30-14:40 Discussion

14:40-15:00 **Amjad Horani, MD**, Washington University School of Medicine, USA, *Novel PCD-Related Genes – Cilia and Beyond*

15:00-15:10 Discussion

15:10-15:30 **Kim Nielsen, MD**, Copenhagen University Hospital, Denmark  
*The Upcoming RCT in BESTCILIA (bestcilia.eu) and the Best Outcome Measures in PCD Trials*

15:30-15:40 Discussion

15:40 – 15:45 **Eitan Kerem MD**, Hadassah Hebrew University Medical Center, Israel  
**Thomas Ferkol, MD**, Washington University School of Medicine, USA  
*Closing Remarks*