



European
Reference
Networks

ERN-LUNG
RARE RESPIRATORY DISEASES

II. INTERNATIONAL ERN LUNG PCD MEETING

6TH - 8TH APRIL 2025
Naples - ITALY

Bringing together scientists from across the world to share advances in Primary Ciliary Dyskinesia diagnosis, clinical research and care.

Programme



Rationale

Primary Ciliary Dyskinesia (PCD) is a rare genetic disorder with significant respiratory implications, affecting both pediatric and adult populations. The condition is primarily characterized by recurrent upper and lower respiratory tract infections, including frequent bronchitis and pneumonia, which, over time, may progress to chronic obstructive pulmonary disease and irreversible respiratory failure. Despite its clinical impact, the prevalence of PCD is underreported, as many patients remain undiagnosed due to variations in diagnostic accessibility and potential underrecognition of the disease itself.

The ERN-LUNG network, through its PCD-specific subgroup, plays a pivotal role in standardizing diagnostic approaches and treatment protocols, thereby improving patient outcomes. The network is actively involved in the development of clinical guidelines and the promotion of innovative therapeutic strategies.

This meeting aims to foster collaboration among experts in the field to advance the understanding of PCD, enhance diagnostic accuracy, and share best practices in treatment. Key objectives include:

- Discussing the latest advancements in diagnostic tools and techniques for PCD.
- Reviewing current treatment protocols and identifying areas for improvement.
- Exploring novel therapeutic approaches and clinical trials.
- Promoting international collaboration to harmonize standards of care and improve patient management globally.

Through these efforts, the congress seeks to drive progress in the clinical management of PCD and contribute to the development of more effective therapies for this challenging condition.



Scientific Committee

Francesca Santamaria

Local Host

Associate Professor of Pediatrics

Dept of Translational Medical Sciences • Federico II University, Naples - Italy

Heymut Omran

ERN-LUNG PCD Core Coordinator and Lead of the International PCD Registry

Professor in Pediatrics

University Hospital and Medical Faculty of the University of Münster, Germany

Kim Nielsen

Director of the PCD-CTN

Professor in Pediatrics and Pediatric Pulmonology

Copenhagen University Hospital, Denmark

Panayiotis Yiallourous

Professor of Pediatrics and Pediatric Pulmonology

Medical School of the University of Cyprus

Johanna Raidt

Pediatric Pulmonologist and Clinical Research Coordinator

University Hospital and Medical Faculty of the University of Münster, Germany

Petra Pennekamp

PhD in Developmental Biology, Research Coordinator

University Hospital and Medical Faculty of the University of Münster, Germany

Panayiotis Kouis

PhD in Environmental Health and Visiting Lecturer

Medical School of the University of Cyprus

Michele Manion

Co-founder and Executive Director PCD Foundation, USA

Pinelopi Anagnostopoulou

Assistant Professor of Pediatrics and Pediatric Pulmonology

Medical School of the University of Cyprus

Melissa Borrelli

Doctor in Pediatric Pulmonology

University Hospital Federico II, Naples - Italy

Faculty

Sara Altaraihi, Denmark

Pinelopi Anagnostopoulou, Cyprus

Ophir Bar-On, Australia

Susanne Böker, Germany

Mieke Boon, Belgium

Melissa Borrelli, Italy

Nicola Brunetti Pierri, Italy

Zuzanna Bukowy-Bieryllo, Poland

Paolo Buonpensiero, Italy

Siobhán Carr, United Kingdom

Adele Corcione, Italy

Maria Elisa Di Cicco, Italy

Gioconda Di Luca, Italy

Valentina Agnese Ferraro, Italy

Giuseppe Fiorentino, Italy

Angelo Florio, Italy

Pietro Formisano, Italy

Brunella Franco, Italy

Giulia Frisso, Italy

Luis J.V. Galletta, Italy

Myrona Goutaki, Switzerland

Eric Haarman, The Netherlands

Rob Hirst, United Kingdom

Amjad Horani, USA

Panayiotis Kouis, Cyprus

Helle Krogh, Denmark

Margaret W. Leigh, USA

Giuseppe Limongelli, Italy

Michael Loebinger, United Kingdom

Niki Tomas Loges, Germany

Bernard Maitre, France

Marcus Mall, Germany

June K. Marthin, Denmark

Pleasantine Mill, United Kingdom

Hannah Mitchison, United Kingdom

Antonio Moreno Galdo, Spain

Jann Mortensen, Denmark

Loretta Müller, Switzerland

Kim Nielsen, Denmark

Heymut Omran, Germany

Jean-François Papon, France

Petra Pennekamp, Germany

Massimo Pifferi, Italy

Dario Prais, Israel

Tavs Qvist, Denmark

Johanna Raidt, Germany

Phil Robinson, Australia

Jobst Röhmle, Germany

Francesca Santamaria, Italy

Adam Shapiro, Canada

Caroline Marie Torp Nygaard, Denmark

Nicola Ullmann, Italy

Sara Valeri, Italy

Woolf Walker, United Kingdom

Panayotis Yiallourous, Cyprus



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Sunday April 6th, 2025

17:00 Registration

18:00 Opening Remarks • *F. Santamaria, G. Limongelli, P. Formisano*

18:30 History of Pompeii • *G. Di Luca*

19:00 **Plenary Lecture**

Ciliopathy patient variants reveal organelle-specific functions for TUBB4B in axonemal microtubules • *P. Mill*

19:30 Welcome reception

08:00 Registration and welcome coffee

09:00 Opening Remarks • *F. Santamaria, N. Ullmann, M. Pifferi, S. Valeri, H. Omran, A. Florio, V. A. Ferraro*

KEYNOTE LECTURE • Chairs: *O. Bar-On, N. Ullmann*

09:20 Effect of inflammatory conditions on the properties of the airway surface fluid.
Implications for mucociliary clearance disorders • *L. J. V. Galletta*

BEST PRACTICE OF CARE FOR PCD • Chairs: *P. Yiallourous, S. Carr*

09:40 What kind of blood tests shall we perform in the regular care of PCD? • *S. Altaraihi, T. Qvist*

10:00 ENT manifestations: presentation and treatment • *M. Goutaki, J. F. Papon*

10:20 Imaging of the lungs (X-ray, CT and MRI) • *P. Robinson, J. Röhmhel*

10:40 Sputum analysis (bacteriology, DNA sequencing) • *H. Krogh, W. Walker*

11:00 Coffee Break

GENETIC AND CLINICAL HETEROGENEITY IN PRIMARY CILIARY DYSKINESIA

Chairs: *G. Frisso, P. Pennekamp*

11:30 OFD1: one gene, several ciliopathies • *B. Franco*

11:45 Jeune syndrome and more (IFT74 related diseases) • *H. Mitchison*

PCD CARE IN NAPLES

12:00 30 years of PCD clinical care and research: my perspective • *F. Santamaria*

RESEARCH AND CLINICAL TRIALS: THE PRESENT AND THE FUTURE

12:15 Results of the AIRLEAF study and updates on the Boehringer Bronchiectasis program • *M. Mall*

ORAL PRESENTATIONS SELECTED FROM ABSTRACTS • Chairs: *M. Borrelli, P. Kouis*

12:35 Role of thoracic magnetic resonance imaging in the follow-up of Primary Ciliary Dyskinesia
G. Bertolucci

12:46 Exhaled volatile organic compounds to differentiate between genetic variants in patients with Primary Ciliary Dyskinesia; Volatile organic compounds in patients with Primary Ciliary Dyskinesia with or without prophylactic azithromycin • *J. Israëls*

12:57 Yield of Genetic test for Primary Ciliary Dyskinesia (PCD) by next-generation sequencing: application to the pediatric population of Southern Italy • *F. Uomo*

13:08 Semi-automated quantification of ciliary beat pattern • *M. Lopes*

13:20 Use of rhDNase in PCD bronchiectasis • *M. Heching*

13:32 Comparison of pulmonary function tests in PCD patients colonized with Haemophilus influenzae and Pseudomonas aeruginosa • *Ö. Faruk İpek*

13:45 Lunch

Poster Walk • *P. Anagnostopoulou, A. Corcione, V. A. Ferraro, M. Goutaki, C. M. Torp Nygaard, P. Pennekamp*

UPDATES ERN-LUNG AND THE ERN-LUNG PCD CORE

15:30 Are official ERN-LUNG PCD Key Performance Indicators for PCD still valid and other aspects
H. Omran

ERN-LUNG PCD REGISTRY STUDIES • Chairs: *E. Haarman, M. W. Leigh*

15:45 Nasal NO measurement in PCD (genotype-phenotype correlations) • *J. Raidt*

16:00 Which patients need diagnostic work-up for PCD? • *H. Omran*

16:15 SNOT scores in PCD • *J. Raidt*

16:30 Non-tuberculosis Mycobacteria in PCD • *D. Prajs*

16:45 Prospective PCD Exacerbations Study • *P. Anagnostopoulou*

17:00 Familial Concordance Study • *P. Kouis*

17:15 **Poster Prizes**

08:00 Welcome coffee

PATIENT PERSPECTIVES AND NATIONAL PERSPECTIVES FOR PCD CARE

Chairs: *M. W. Leigh, S. Böker*

Issues to be addressed from the national perspective:

- What are your urgent needs to improve clinical care?
- What are your urgent research topics for PCD?
- What do you expect from the ERN-LUNG PCD Core?

08:30 The Italian PCD perspective • *N. Ullmann*

08:45 The Spanish Registry within the International ERN-LUNG PCD Registry

A. Moreno Galdo

09:00 The British perspective • *W. Walker*

09:15 The US perspective and Canadian Perspective, US PCD Foundation Activities

A. Shapiro

09:45 *Coffee Break*

10:15 **ERN-LUNG PCD-CTN: Welcome and Update** • *K. G. Nielsen*

RESEARCH AND CLINICAL TRIALS: THE PRESENT AND THE FUTURE

Chairs: *E. Haarman, M. W. Leigh*

10:30 Gene therapy for genetic disorders with AAV vectors targeting the liver

N. Brunetti-Pierri

10:50 Update of the Phase I trial DNAI mRNA therapy (UK, DK, DE) and how to get ready for genotype-specific trials in PCD • *M. Loebinger, K. G. Nielsen*

11:30 Brensocatib for Bronchiectasis is on the way to the patient – what does this mean for patients with PCD? • *J. Röhm*

11:50 Pulmonary radioaerosol mucociliary clearance assessment • *J. Mortensen*

AIRWAY CLEARANCE TECHNIQUES

12:15 Latest advice on airway clearance techniques • *P. Buonpensiero*

12:30 *Lunch*

DIAGNOSIS AND MANAGEMENT OF PCD FOR EARLY CAREER CLINICIAN SCIENTISTS

Chairs: *G. Fiorentino*

14:00 The role of nasal nitrite oxide measurement for the diagnosis of PCD

J.K. Marthin, M. Boon

14:20 The role of TEM and IF for the diagnosis of PCD • *Z. Bukowy-Bieryllo, N.T. Loges*

14:40 The role of High-Speed Videomicroscopy and in vitro ciliogenesis for the diagnosis of PCD

L. Müller, R. Hirst

15:00 The role of Genetic testing in PCD • *A. Horani, H. Omran*

15:20 Management of PCD • *M. E. Di Cicco, B. Maitre*

15:40 **Closing remarks** • *F. Santamaria, K. G. Nielsen, H. Omran, M. W. Leigh, S. Valeri*

UNDER THE PATRONAGE OF:



WITH THE CONTRIBUTION OF:





VENUE

University of Naples “Federico II” Congress Centre

Via Partenope, 36 - 80121

INFO

Participation in the conference is free of charge. Registration includes: access to the scientific sessions, the conference kit, lunches, coffee breaks and a certificate of attendance.

ONLINE STREAMING

The meeting will be broadcast live via online streaming, allowing participants to join remotely from anywhere. To access the streaming link and detailed connection instructions, please visit the official event website: <https://pcdnaples2025.centercongressi.com/home.php>. Navigate to the Info section, where you will find all necessary access links and further guidelines to facilitate your connection.