AUGUST 2023

BEAT-PCD
BETTER EXPERIMENTAL APPROACHES TO TREAT PCD

ISSUE 7

Newsletter

In this issue of the newsletter you will find details of the Annual PCD Research Meeting 2023, PCD-related content at the ERS Congress 2023, newly formed slack workspace for PCD diagnosis and ECR-PCD Expert Talk Series.

Agenda for Annual PCD Research Meeting 2023

You can register for the in-person Annual PCD Research Meeting happening at the University of Milano on September 8th 2023 here.

Morning training program:

9.00 Welcome and introduction - Chairs: Myrona Goutaki, Heymut Omran
Maria Francesca Patria (Italian Representative),
Amelia Shoemark, Myrona Goutaki (BEAT-PCD representatives),
Johanna Raidt, Heymut Omran (ERN-LUNG representatives)

9.10 Patient perspective: Italian patient representative -
(Associazione italiana discinesia ciliare primaria - sindrome die Kartagener Aps)

9.20 Patient perspective: BEAT-PCD patient representative - Lucy Dixon (PCD Support UK)

9.30 Italian Perspective - Nicola Ullmann

9.45 Training talks/interactive session - Chairs: Rob Hirst, Nicola Ullman

- Assessment of human airway ciliary beating for the diagnosis of Primary Ciliary Dyskinesia - Mathieu Bottier (20min inc. questions)
- Panel discussion: What's needed for multi-disciplinary management in PCD?
  Open discussion with a multidisciplinary panel of patient representatives and healthcare professionals involved in the care of people with PCD (25min discussion)

10.30 Research highlights from selected early career member abstracts -
Chairs: Katie Horton, Yin Ting Lam
This session will provide a platform for people with ERS posters to give short oral presentations of their work (5min each followed by 15min joint discussion)

- Variability of lung function in patients with primary ciliary dyskinesia (PCD): longitudinal analysis from the PROVALF-PCD cohort - Bruna Rubbo
- Assessing personalised airway clearance techniques in Primary Ciliary Dyskinesia with 129Xe ventilation MRIXX - Lynne Schofield
- Evaluation of sleep disordered breathing and related factors in primary ciliary dyskinesia patients - Nagehan Emiralioglu
- Infertility among adults with primary ciliary dyskinesia - Leonie Schreck
- The importance of collaboration with expert centers on PCD diagnosis in limited-resource countries - Mine Kalyoncu

Morning training program (continued):

11.10 Coffee break

11.30 Difficult diagnosis and management cases presentations -
Chairs: Amjad Horani, Panayiotis Kouis
Difficult cases (4 cases x10min + 5min discussion each)

- CFAP74 mutations or NEK10 mutations - Luisa Biebach
- Combined approaches including long-read sequencing address the diagnostic challenge of HYDIN in primary ciliary dyskinesia – Debbie Morris-Rosendahl
- More tbd – You are invited to contact us if you would like to present a difficult case.

12.30 Lunch

Afternoon European PCD meeting:

13.30 Plenary Talk - Chairs: Claudia Kuehni, Kim Nielsen
The challenges of monitoring disease in people with PCD - Jane Lucas (30min inc. discussion)

14.00 Nasal nitric oxide measurement in children for the diagnosis of PCD: ERS technical standard - Nicole Beydon (15mins + 5min discussion)

14.20 Clinical trials updates - Chairs: Felix Ringshausen, Phil Robinson
(15min + 5min discussion each)

- ReCode Therapeutics Update - John Matthews
- Ethris GmbH Update - Thomas Langenickel
- Mucociliary Clearance Assays - Pulmonary Radioaerosol Mucociliary Clearance - June Marthin
- CLEAN-PCD trial results - Felix Ringshausen

15.40 Coffee break

16.10 Update on European and international PCD projects -
Chairs: Amelia Shoemark, Petra Pennekamp (15min each including questions)

- Update on the US PCD network - Tom Ferkol
- Update on the ERN-LUNG PCD Clinical Trials Network - Kim Nielsen
- Update on the ERN-LUNG PCD international PCD registry and audits - Johanna Raidt
- Update on BEAT-PCD - Myrona Goutaki
- Plans for next 3 years of BEAT-PCD - Amelia Shoemark

17.30 Meeting close and summary

18.00 Get together with some snacks and drinks

PCD-related content at the ERS Congress 2023

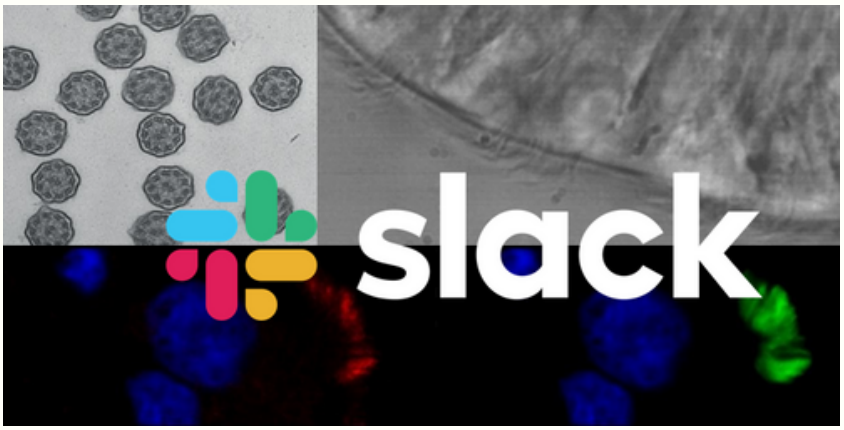
Sunday 10th September

08.00-09.30	PS-14	**Bronchiectasis registries, cohort and subgroups** A complete test panel avoids missed PCD diagnosis in adults with mild clinical phenotype *D. Calmes* The Royal Brompton hospital adult primary ciliary dyskinesia (PCD) service: 2 years on *S. Peake*
	PS-19	**Seeing is believing: new interventions in respiratory education** An assessment of the information and educational needs of patients and caregivers of people with primary ciliary dyskinesia: an ERS clinical research collaboration *J. Röhmel*
10:15-11:45	SILVER	**ALERT 1: pharmacological treatments** Safety and efficacy of idrevloride in people with primary ciliary dyskinesia: a double-blind, randomized, placebo-controlled crossover trial (CLEAN-PCD) *T. Ferkol*
11.45-13.20	STUDIO (WHITE)	**Paediatric year in review** Primary ciliary dyskinesia: diagnosis and management *A. Shoemark*
12.30-14.00	PS-16	**Basic science of respiratory infection** An exploratory study highlighting the use of night-time cough frequency as a potential marker of disease surveillance in patients with Primary ciliary dyskinesia and Cystic fibrosis *E. Robson*
15.45-17.00	BROWN 1+2	**Technical and epidemiological novelties in paediatric lung function and sleep** Nasal nitric oxide measurement in children for the diagnosis of primary ciliary dyskinesis: ERS technical standard *N. Beydon*
16.00-17.30	PS-3	**New insights and biomarkers in chronic obstructive pulmonary disease and epithelial disorders** A new method to estimate human bronchial epithelial cells ciliary beat frequency *S. Palea* Combined approaches including long-read sequencing address the diagnostic challenge of HYDIN in primary ciliary dyskinesia *D. Morris* Increasing the diagnostic yield in Primary Ciliary Dyskinesia via whole gene sequencing *L. Briggs* Knocking-down FAM13A during lung epithelial differentiation alters the development of cilia *L. Yrlid* Primary cilium (PC) associated BBSome gene 1, 7 and 9 expression during the 2D and 3D culture of mesothelial cells under oxidative stress *R. Jagirdar* The utility of RNA and transcriptome analysis in improving diagnostics and understanding disease mechanism in Primary Ciliary Dyskinesia *J. Legebeke* Variants in CEP164 cause PCD: expanding the spectrum of primary and motile ciliopathy overlap *J. Coles*
	PS-4	**Cutting-edge approaches to model and treat lung disease** Development of a Xenopus-based assay for high-throughput evaluation of mucociliary flow *M. Chatzifrangkeskou* Establishing the Conditionally Reprogrammed Cell Technology for precision medicine in PCD *R. Urbantat* Proteomic profiling of air-liquid-interface (ALI)-cultured primary ciliary dyskinesia (PCD) nasal epithelia *K. Horton*
16.00-17.30	PS-5	**Sleep-disordered breathing in other respiratory diseases and nocturnal hypoventilation** Evaluation of sleep disordered breathing and related factors in primary ciliary dyskinesia patients *N. Emiralioğlu*
	PS-12	**Paediatric bronchology: new insights** Alexithymia in primary ciliary dyskinesia *G. Unal*

Monday 11th September		
08.00-09.30	PS-20	**Deep learning, new technologies and biomarkers in imaging** PCD-AID: artificial intelligence diagnosis of primary ciliary dyskinesia *M. Bottier* Utilising computer vision artificial intelligence to identify defects in airway ciliary motility and mucociliary clearance *M. Bottier*
12.30-14.00	PS-10	**Translational model in chronic airway disease** A survey of patient engagement in primary ciliary dyskinesia *C. Webster* Extracellular vesicles from PBDE-47 treated M(LPS) THP-1 macrophages modulate gene expression in ALI culture of airway epithelium *M. Profita*
12.30-14.00	PS-13	**Epidemiology of rare paediatric respiratory diseases** A BEAT-PCD consensus statement: a core outcome set for pulmonary disease interventions in primary ciliary dyskinesia *E. Haarman* Association between upper and lower respiratory disease in primary ciliary dyskinesia *YT. Lam* Evaluating gene-disease relationships in motile ciliopathies: an international ClinGen and BEAT-PCD ERS CRC collaboration *S. Crowley* Fertility counselling in people with primary ciliary dyskinesia *L. Schreck* Genotype-phenotype associations in primary ciliary dyskinesia *E. Pedersen*
12.30-14.00	PS-13	Lung function and airway microbiology in primary ciliary dyskinesia (PCD): cross-sectional analysis from the PROVALF-PCD cohort, a BEAT-PCD Collaboration *B. Rubbo* Neonatal problems of people with primary ciliary dyskinesia in Switzerland *M . Goutaki* Physical activity in people with primary ciliary dyskinesia *E. Pedersen* Piloting the FOLLOW-PCD questionnaire: a clinical and research tool *M . Goutaki* Variability of lung function in patients with primary ciliary dyskinesia (PCD): longitudinal analysis from the PROVALF-PCD cohort, a BEAT-PCD Collaboration *R. Rubbo*
12.30-14.00	PS-14	**Nursing and digital health** Adult Primary Ciliary Dyskinesia (PCD) physiotherapy standards of care in England *E. Shepherd*
12.30-14.00	PS-17	**Bronchiectasis treatments** Retrospective review of treatment approaches for Pseudomonas aeruginosa infections in patients with Primary Ciliary Dyskinesia: Insights from a Danish cohort *M. Holgersen*
16.00-17.30	PS-13	**Poster session: Effects of CFTR modulator therapy in cystic fibrosis** Infertility among adults with primary ciliary dyskinesia *L. Schreck*

Tuesday 12th September		
08.00-09.30	PS-2	**Novelties in lung function testing** Lung clearance index as a marker of lung functional impairment in primary ciliary dyskinesia: preliminary results of a systematic review of the literature *A. Matthaiou*
	PS-20	**Ongoing imaging-related clinical studies in chronic obstructive pulmonary disease, asthma, interstitial lung disease and other pulmonary diseases** Multisite Hyperpolarized 129Xe MRI Study of Pediatric Primary Ciliary Dyskinesia *W. Wee*
09.30-10.45	YELLOW 1+2+3	**New basic insights into the mechanisms of acute and chronic lung diseases** A primary ciliary dyskinesia model of static cilia for research application *C. Jackson*
12.30-14.00	PS-13	**Non-cystic fibrosis bronchiectasis and other chronic lung infections in children** Disease manifestations in siblings with primary ciliary dyskinesia *D. Gatt* Does timing of diagnosis effect outcome in primary ciliary dyskinesia? *D. Gatt* Nasal Mucociliary Clearance of 99mTc-albumin colloid: a quantitative test of nasal ciliary function and potential outcome parameter in primary ciliary dyskinesia *J. Marthin* Genomics of Haemophilus influenzae strains in a paediatric Primary Ciliary Dyskinesia cohort *K. Schmidthaler* Patterns of E-cigarette Usage Among Adolescents with Primary Ciliary Dyskinesia and Cystic Fibrosis *D. Gatt* Repeating ciliary videomicroscopy improves the specificity for PCD diagnosis *L. Benchimol* Use of Immunofluorescence Staining in Primary Ciliary Dyskinesia Diagnosis *M. Kalyoncu*
13.45-15.15	PURPLE	**Genetics in paediatric respiratory diseases** Models for ciliated airway epithelial cells and personalised medicine *A. Berical*
16.00-17.30	PS-10	**Cystic fibrosis: disease monitoring, biomarkers, and psychosocial issues** Exploring the attitudes and knowledge of the caregivers of children with cystic fibrosis and primary ciliary dyskinesia regarding COVID-19 vaccination *P. Ergenekon*
	PS-12	**Airway clearance, physical activity and exercise in children and adults with respiratory disorders** Assessing personalised airway clearance techniques in Primary Ciliary Dyskinesia with 129Xe ventilation MRI *L. Schofield* Impaired muscle oxygenation, exercise capacity, and activities of daily living in primary ciliary dyskinesia with inspiratory muscle weakness *H. Sonbahar Ulu* Inspiratory muscle strength is an independent predictor for six-minute walk distance in primary ciliary dyskinesia *H. Sonbahar Ulu*
Wednesday 13th September		
09:45-12:45	BROWN 3	**How to build a successful, sustainable, international network** *Poster by BEAT-PCD* Getting involved in international research networks – patient and caregiver perspectives *L. Dixon*

Join the newly formed slack workspace for PCD diagnosis!



The "**Work Package 7 - Improving PCD Diagnosis**" has created a slack workspace to provide a collaborative platform about PCD diagnosis. In specific channels, BEAT-PCD members will have the opportunity to ask questions about protocols and methods and to share difficult cases (TEM, HSVM and IF).

Slack allows the sharing of images, videos or PDF files on the platform. The platform will be monitored and moderated at least twice a week by WP7 members providing answers or resources. We are asking members to keep opinion pieces to the minimum and we encourage the exchange of only evidence-based information.

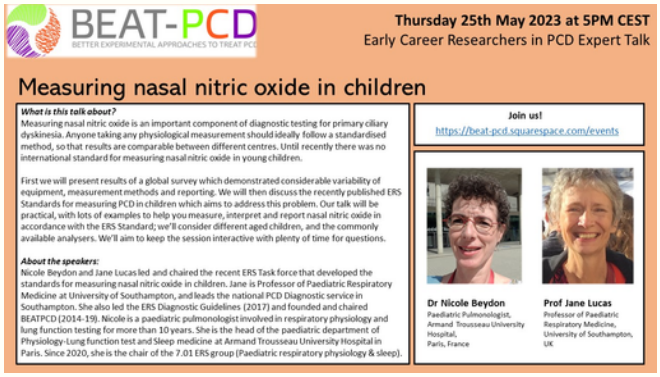
If you would like to join, please contact Dr Mathieu Bottier (m.bottier@rbht.nhs.uk) to receive an invite.

ECR-PCD Expert Talk Series

SAVE THE DATE:
Our next session will be on **Wednesday 4th October 2023** at **17.00 CEST.** We are super excited to be having Professor Hannah Mitchison and Dr Myrona Goutaki talk about **"Genotype-Phenotype associations in PCD".**

During our last expert session, Dr Nicole Beydon and Professor Jane Lucas talked in depth about the recently published ERS Technical Standard.

Catch up on the session and all of our previous expert sessions here.


BEAT-PCD
BETTER EXPERIMENTAL APPROACHES TO TREAT PCD
Thursday 25th May 2023 at 5PM CEST
Early Career Researchers in PCD Expert Talk
Measuring nasal nitric oxide in children
What is this talk about?
Measuring nasal nitric oxide is an important component of diagnostic testing for primary ciliary dyskinesia. Anyone taking any physiological measurement should ideally follow a standardised method, so that results are comparable between different centres. Until recently there was no international standard for measuring nasal nitric oxide in young children.
First we will present results of a global survey which demonstrated considerable variability of equipment, measurement methods and reporting. We will then discuss the recently published ERS Standards for measuring PCD in children which aims to address this problem. Our talk will be practical, with lots of examples to help you measure, interpret and report nasal nitric oxide in accordance with the ERS Standard; we'll consider different aged children, and the commonly available analysers. We'll aim to keep the session interactive with plenty of time for questions.
About the speakers:
Nicole Beydon and Jane Lucas led and chaired the recent ERS Task force that developed the standards for measuring nasal nitric oxide in children. Jane is Professor of Paediatric Respiratory Medicine at University of Southampton, and leads the national PCD Diagnostic service in Southampton. She also led the ERS Diagnostic Guidelines (2017) and founded and chaired BEATPCD (2014-19). Nicole is a paediatric pulmonologist involved in respiratory physiology and lung function testing for more than 10 years. She is the head of the paediatric department of Physiology-Lung function test and Sleep medicine at Armand Trousseau University Hospital in Paris. Since 2020, she is the chair of the 7.01 ERS group (Paediatric respiratory physiology & sleep).
Join us!
https://beat-pcd.squarespace.com/events
Dr Nicole Beydon
Paediatric Pulmonologist, Armand Trousseau University Hospital, Paris, France
Prof Jane Lucas
Professor of Paediatric Respiratory Medicine, University of Southampton, UK

Ways to FIND and JOIN us!

Help us continue to expand our network and work collaboratively by:
Connecting on Twitter @beatpcd Friending on Facebook www.facebook/beatpcd
Keeping updated with PCD news by joining the BEAT-PCD mailing list (email BEATPCD@ersnet.org)
Joining the Early Career Researchers mailing list (email K.L.Horton@soton.ac.uk)
Visiting our website www.beat-pcd.squarespace.com