MARCH 2024

BEAT-PCD
BETTER EXPERIMENTAL APPROACHES TO TREAT PCD

ISSUE 8

Newsletter

Welcome to issue 8 of the BEAT-PCD newsletter. This edition is an update and summary of the recent activities of BEAT-PCD. We include links to catch up on anything you missed and opportunities to get more involved in our work, including upcoming meetings and projects.

Update on BEAT-PCD Projects

PCD bioresources & biomarkers

We are developing a European PCD biobank. We have begun with a mapping exercise of existing biobanks and are looking for individuals interested in taking part. We recently distributed a survey to all stakeholders about current biobanking and bioresources for PCD. Our first biobank project will be looking at disease biomarkers and are finalising systematic review on biomarkers for PCD, soon to be submitted for publication.

'complete:PCD' - If you see patients with PCD and are interested to receive more information or to take part please complete this short survey

Open access PCD database

CiliaVar, the online database containing all known PCD variants, with the aim of collecting clinical and diagnostic data to improve PCD diagnosis and support patient stratification and eligibility for clinical trials. To date, more than 2000 variants have been included, and in the near future, the hope is to test-run the five most common genes DNAH5, DNAH11, CCDC39, CCDC40, DNAI1.

A spin-off project is a collaboration with ClinGen, an NIH-funded resource dedicated to defining the clinical relevance of genes and variants. An international panel of experts from BEAT-PCD and NIH has been assembled to curate disease-gene relationships in motile ciliopathies; of 98 identified motile ciliopathy genes, 57 have to date been curated. Further, a variant panel of experts has also been established, tasked with assessing the pathogenicity of known variants.

Patient engagement activities

Almost 400 patients and parents of children with PCD participated in worldwide survey about **patients' research priorities in PCD** (manuscript submitted soon)

With help of participants, COVID-PCD study was renamed to **Living with PCD** study to reflect that the current focus is any research question relevant for people with PCD (new website and email). Latest newsletter to study participants. In summer 2024, there will be a study re-launch and new wave of recruitment.

Latest publications:

- Incidence and severity of SARS-CoV-2 infections in people with primary ciliary dyskinesia
- Diagnostic testing in people with primary ciliary dyskinesia: an international participatory study
- Fertility care among people with primary ciliary dyskinesia
- Genotypes and associations with symptoms in primary ciliary dyskinesia (under review)

Further manuscripts on infertility and physical activity will be submitted soon.

Neglected areas for PCD research

Focus on upper airways

Latest manuscripts from the EPIC-PCD study:

- Lack of Correlation of Sinonasal and Otologic Reported Symptoms With Objective Measurements Among Patients With Primary Ciliary Dyskinesia
- Association between upper and lower respiratory disease in primary ciliary dyskinesia

A phenotype-genotype association study on otologic disease has started, including a longitunidal analysis of hearing in Bicetre (Paris, France) cohort of adult PCD patients.

Improving PCD diagnosis

- An nNO patient/carer information sheet based on content from the published technical standard is being developed.
- Training package for high-speed video microscopy, with media supplied within the UK PCD service (courtesy Prof O'Callaghan) is also in development. We will be asking BEAT-PCD members to participate going forward.
- Systematic review on automated /artificial intelligence/machine learning-led/computerised approaches to PCD diagnostics
- BEAT-PCD supports the ongoing ERS/ATS Task Force for the development of a revised joint diagnostic guideline.

Clinical standardisation & education

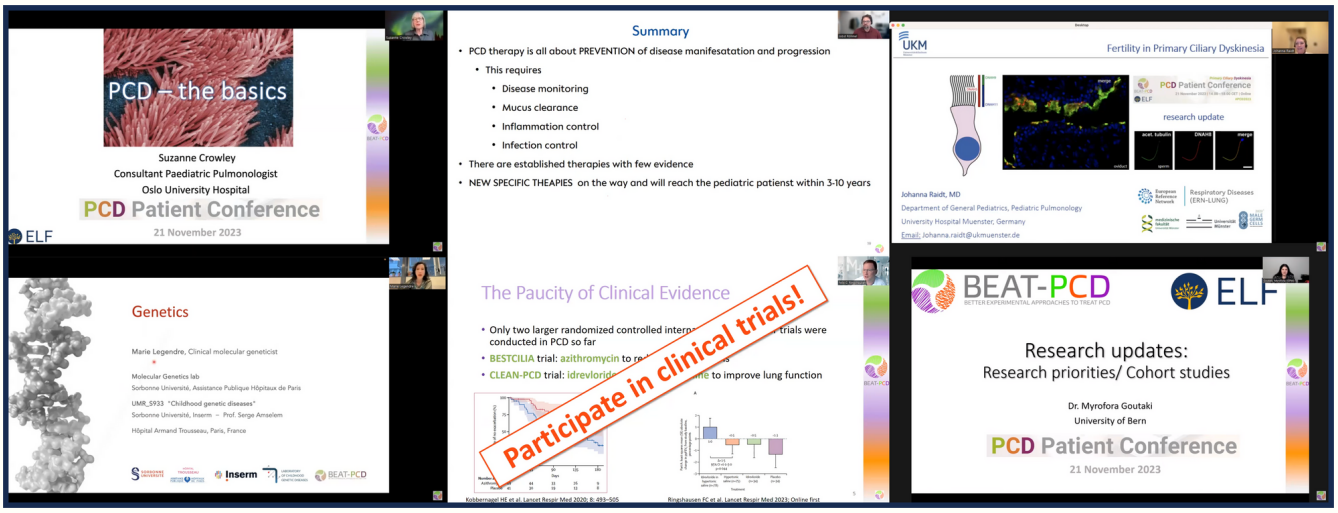
- Pilot data analysis and ongoing cognitive validation to refine FOLLOW-PCD patient questionnaire. Currently available in 3 age-specific versions, 11 languages
- FOLLOW-PCD ENT clinical examination module
  - Ongoing Delphi process to refine the module
- Standardised form for transition of people with PCD to adult care developed and currently piloted
- Consensus statement on routine blood testing in PCD is in development.

Clinical outcome measures

An international panel of specialists involved in PCD care and patients with PCD developed a **consensus definition of ENT exacerbations** for children and adults with PCD, for use in clinical research (manuscript submitted).

Published: [A BEAT-PCD consensus statement: a core outcome set for pulmonary disease interventions in primary ciliary dyskinesia]

Feedback from 2nd International PCD Patient Conference 2023



300 registered participants from over 12 countries, with over 100 participants connected at all times.

Highlight most successful session:

Living with PCD session organised completely by patients themselves

When asked what aspects of the conference did you find most valuable, some responded:

"explanation of PCD & the 'tips & tricks' how to treat PCD and live with it - especially in the child presentation."

"the themes presented, the progress of the presentations, the testimonies of parents"

Recordings can be accessed here

PCD On The Move Conference 2024

Represented by both BEAT-PCD co-chairs and several members of the management committee, BEAT-PCD participated actively at the **"PCD On The Move Conference 2024"** organised by the PCD Foundation in Puerto Rico in January 25th-28th.

Our members participated with lectures, chaired sessions, organised break-out trainings, exchanged opinions and ideas for future collaborations, with **more than 200 colleagues** from North and South America and all over the world. The project **"Infertility among adults with primary ciliary dyskinesia"** from the **Living with PCD study**, a large participatory study of the BEAT-PCD network hosted at the University of Bern, received the prize for the **Best Clinical Research Poster presentation**.







ECR-PCD webinars continue successfully

Watch webinar recordings here

Measuring nasal nitric oxide in children
Genotype-phenotype associations in PCD
Fertility in PCD
Paediatric PCD clinical cases

Next meetings & opportunities to get involved

Register here for next ECR-PCD Symposium: 17th April 2024 5:30PM CEST "Adult Clinical PCD Cases"

Look forward to interactive cases discussions, moderated by Natalie Lorent MD PhD (Leuven, Belgium)

Register here for

WORLD BRONCHIECTASIS CONFERENCE
Dundee, Scotland
4 - 6 July 2024

Half-day programme dedicated to PCD organised by BEAT-PCD

Abstract submission open here

European Reference Networks
ERN-LUNG
RARE RESPIRATORY DISEASES

INTERNATIONAL ERN LUNG-PCD MEETING

APRIL 10 - 12 2024

NICOSIA, CYPRUS

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

Organizers:
University of Münster, University of Cyprus

Registration & Contact Info

Register here

Join slack for PCD diagnosis!

Ask questions about protocols and methods and share difficult cases (TEM, HSVM and IF). We are encouraging the exchange of only evidence-based information. **To join**, contact Dr Mathieu Bottier (**m.bottier@rbht.nhs.uk**).

FIND US

Join BEAT-PCD mailing list

Join early career researchers' mailing list

BEAT-PCD
BETTER EXPERIMENTAL APPROACHES TO TREAT PCD

European Reference Network | ERN-LUNG
European Reference Network for rare respiratory diseases

PCD Annual Research Meeting

SAVE THE DATE

VIENNA
AUSTRIA

6TH SEPTEMBER
2024

- Stay up to date on research, therapies, and collaborations related to PCD
- Get involved in ongoing projects, and contribute to setting up the goals for the next period
- Discuss difficult diagnostic and clinical PCD cases
- Consultation on the joint ERS/ATS diagnostic guidelines and management consensus guidelines
- Early career member research prizes

Scientific and organising secretariat

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