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We want to hear from you

We value the opinion of our readers and we are looking forward to receive your feedback about this newsletter.

Please send us a personal email with your suggestions or comments to the following address:

Attn: Stephanie Gallant
BEATPCD@ersnet.org



The BEAT-PCD Clinical Research Collaboration (CRC) is funded by the European Respiratory Society.

ISSUE 2 | JUNE 2021

BEAT-PCD NEWSLETTER



ANNUAL EVENTS

Online BEAT-PCD training school 2021

September 4th 2021

13:00 CET/12.00 UK

Online BEAT-PCD annual meeting 2021

September 9th 2021

13:00 CET/ 12.00 UK



*Registrations for both events, agendas and further information
will be made available on our website:*

<http://beat-pcd.squarespace.com/>

The BEAT-PCD annual events take place on either side of ERS international congress which for 2021 will be a virtual event. Additional info for the ERS virtual event is provided [here](#).





Message from the BEAT PCD chairs

Welcome to the 2nd edition of the BEAT-PCD newsletter.

This edition focuses on the progress achieved during the first year of our project. Overall, we are happy to report that despite the difficulties caused by the COVID-19 pandemic, the BEAT-PCD Clinical Research collaboration was able to achieve some important milestones and set the pace for the years to follow.

In addition, you can read about recent scientific publications from collaborative projects focusing on improving diagnosis and treatment of PCD including the BEAT-PCD editorial published in the European Respiratory Journal. Last but not least, you can find more about the Early Career Researchers in PCD (ECR-PCD) Expert Talks series, one of the most exciting training initiatives of BEAT-PCD.

We hope that you will enjoy reading this edition.

Amelia & Myrona

BEAT-PCD 1st year summary

At the end of March 2021, the BEAT-PCD ERS CRC completed successfully its first year. Here, we would like to summarize the progress of our main projects and major achievements, presented by work package (WP).

WP1: Project management and governance

During this year, the BEAT-PCD management committee met regularly to plan and set long and short-term goals. Meetings with our advisory board helped to ensure that all activities of the network are beneficial to all stakeholders and complimentary to the activities of other relevant initiatives. In addition, we hold several work package or project specific meetings with participation of several members of the network as well as representatives of PCD support groups.

WP2: Network of PCD research databases and collaborations

A main project of this work package is 'Ciliavar', an online open database registering gene mutations and specific combinations of disease-causing variants for PCD. We formalized a minimum dataset and already began to populate the database starting with > 1000 PCD cases from the published literature.

WP2 supported the further development and collaboration between existing research databases and networks. Overall, existing research databases increased in patient numbers and quality of contributed data was improved. We analysed and published data providing evidence on disease progression and phenotype-genotype correlations. In collaboration with EMBARC (the bronchiectasis CRC) we extracted and analysed longitudinal data from >500 PCD cases in the international EMBARC bronchiectasis registry. Finally, a new ENT-focused PCD cohort study (EPIC-PCD) aiming to characterise upper airway disease in children and adults with PCD began recruitment in 10 countries and has already recruited >200 patients.

BEAT-PCD 1st year summary

WP3: Patient engagement activities

WP3 was responsible for a series of patient engagement activities. We identified and contacted existing national PCD support groups and in collaboration with the European Lung Foundation (ELF) we held regular meetings with patients to ensure that all our activities addressed patient's needs and priorities. We developed a comprehensive survey to identify gaps in educational material for patients with PCD, which was informed by a literature review and feedback from patients/parents and clinicians. The survey will be translated and circulated widely this year. In addition to our initial aims, we developed COVID-PCD, a cohort study in collaboration with PCD support groups, studying the effects of SARS CoV2 on people with PCD. Results are published regularly on the study website <https://covid19pcd.ispm.ch/> and two manuscripts (study protocol and an original paper on incidence of COVID19 in PCD patients) have been published.



ELF EUROPEAN
LUNG
FOUNDATION

WP4: Clinical trials

WP4 worked towards the development of a PCD clinical trials network (PCD-CTN). The PCD-CTN was established in the framework of the ERN-LUNG and consists currently of 18 trial sites in Europe, which manage in total more than 2000 children and adults with PCD. An important focus of WP4 is identifying and developing standardised and validated outcome measures for clinical trials. We have submitted for publication a scoping review on pulmonary outcome measures in PCD and completed the translation of the QoL-PCD in 5 further languages (Czech, Norwegian, Hebrew, Polish and Swedish). We also encouraged the participation of more centres to the international PCD registry through the network and CRC website.

WP5: Improvement of PCD diagnosis

WP5 established a network of clinicians to combine the expertise of several centres to improve diagnosis of PCD cases and support diagnosis in centres with limited resources. We held our first difficult diagnostic case meeting online on November 5th with 100 healthcare workers attending and we plan the 2nd meeting this summer. WP5 is also associated with the ongoing ERS TaskForce that is developing a "Technical standard on nasal nitric oxide measurement in children for the diagnosis of PCD".

WP6: Improvement of clinical care

We began piloting FOLLOW-PCD (particularly the patient questionnaire part) in clinics and research through several projects e.g. EPIC-PCD, COVID-PCD. We translated the FOLLOW-PCD patient questionnaire in 6 more languages, thus it is now available in 9 languages: English, German, Greek, French, Spanish (also Latin America versions), Dutch/Flemish, Turkish, Danish and Norwegian. In close collaboration with our group of Early Career Researchers participating in BEAT-PCD, we developed a program of training sessions to expand their knowledge in the field of PCD and also to help them acquire transferable skills that will assist them to develop their careers.

WP7: Engagement with the ERS and dissemination of CRC activities and results

We held a successful kick off meeting adjacent to the virtual ERS congress with >200 participants and launched the new revised BEAT-PCD website (www.beat-pcd.squarespace.com) and twitter account (@beatpcd) and a new Facebook group where we regularly post about the CRC ongoing and planned activities and achievements. To increase awareness of PCD and the ERS CRC network we published an editorial about BEAT-PCD in the ERJ and submitted >10 abstracts to the 2021 ERS congress.

You can read more about the objectives and activities of each WP on our website https://beat-pcd.squarespace.com/working-groups-summary_

Featured Research

BEAT-PCD groups have been busy over the past year completing collaborative projects to improve diagnosis and treatment for patients with PCD. Here we highlight some recent publications from the group:

COVID-PCD – a participatory research study on the impact of COVID-19 in people with Primary Ciliary Dyskinesia

Eva SL Pedersen, Eugénie NR Collaud, Rebeca Mozun, Cristina Ardura-Garcia, Yin Ting Lam, Amanda Harris, Jane S Lucas, Fiona Copeland, Michele Manion, Bernhard Rindlishbacher, Hansruedi Silberschmidt, COVID-PCD patient advisory group, Myrofora Goutaki, Claudia E Kuehni

This article describes the BEAT-PCD COVID study which is a longitudinal participatory study that follows people with PCD during the COVID-19 pandemic through online anonymous questionnaires. Results are published in real-time on the study website (www.covid19pcd.ispm.ch) including number of people infected with SARS-CoV-2, severity of disease, and number of people vaccinated against COVID-19. The COVID-PCD study is conducted in close collaboration with people who have PCD and provides a platform for communication between patients, physicians, and researchers. You can read the published manuscript [here](#).

Topological data analysis reveals genotype-phenotype relationships in primary ciliary dyskinesia

Amelia Shoemark, Bruna Rubbo, Marie Legendre, Mahmood R. Fassad, Eric G. Haarman, Sunayna Best, Irma C.M. Bon, Joost Brandsma, Pierre-Regis Burgel, Gunnar Carlsson, Siobhan Carr, Mary Carroll, Matt Edwards, Estelle Escudier, Isabelle Honoré, David Hunt, Gregory Jouvion, Michel Loebinger, Bernard Maitre, Deborah Morris-Rosendahl, Jean-Francois Papon, Camille M. Parsons, Mitali P. Patel, Simon N Thomas, Guillaume Thouvenin, Woolf T. Walker, Robert Wilson, Claire Hogg, HannahM. Mitchison, Jane S. Lucas

Published earlier this year this multinational genotype-phenotype study in PCD describes worse lung function in patients with CCDC39 mutations compared to other patients. Topological data analysis methods are used to show that people with DNAH11 mutations have less neonatal respiratory distress and better FEV1 compared to other patients with PCD. You can read the published manuscript [here](#).

Access to medicines for rare diseases: beating the drum for primary ciliary dyskinesia

Suzanne Crowley, Inês Azevedo, Mieke Boon, Andrew Bush, Ernst Eber, Eric Haarman, Bulent Karadag, Karsten Kötz, Margaret Leigh, Antonio Moreno-Galdó, Huda Mussaffi, Kim G. Nielsen, Heymut Omran, Jean-François Papon, Petr Pohunek, Kostas Priftis, Bernhard Rindlishbacher, Francesca Santamaria, Arunas Valiulis, Michal Witt, Panayiotis Yiallourous, Zorica Zivkovic, Claudia E. Kuehni, Jane S. Lucas for BEAT-PCD

This article was published in ERJ open on behalf of BEAT-PCD in September 2020. Clinical experts from multiple countries highlight the challenges faced for access to medicines in rare diseases such as PCD. The article describes the lack of evidence base for treatments of PCD and proposes the following solutions 1) forming a PCD European clinical trial network to address this situation and 2) conducting n-of-1 trials to access medication. You can read the published manuscript [here](#).

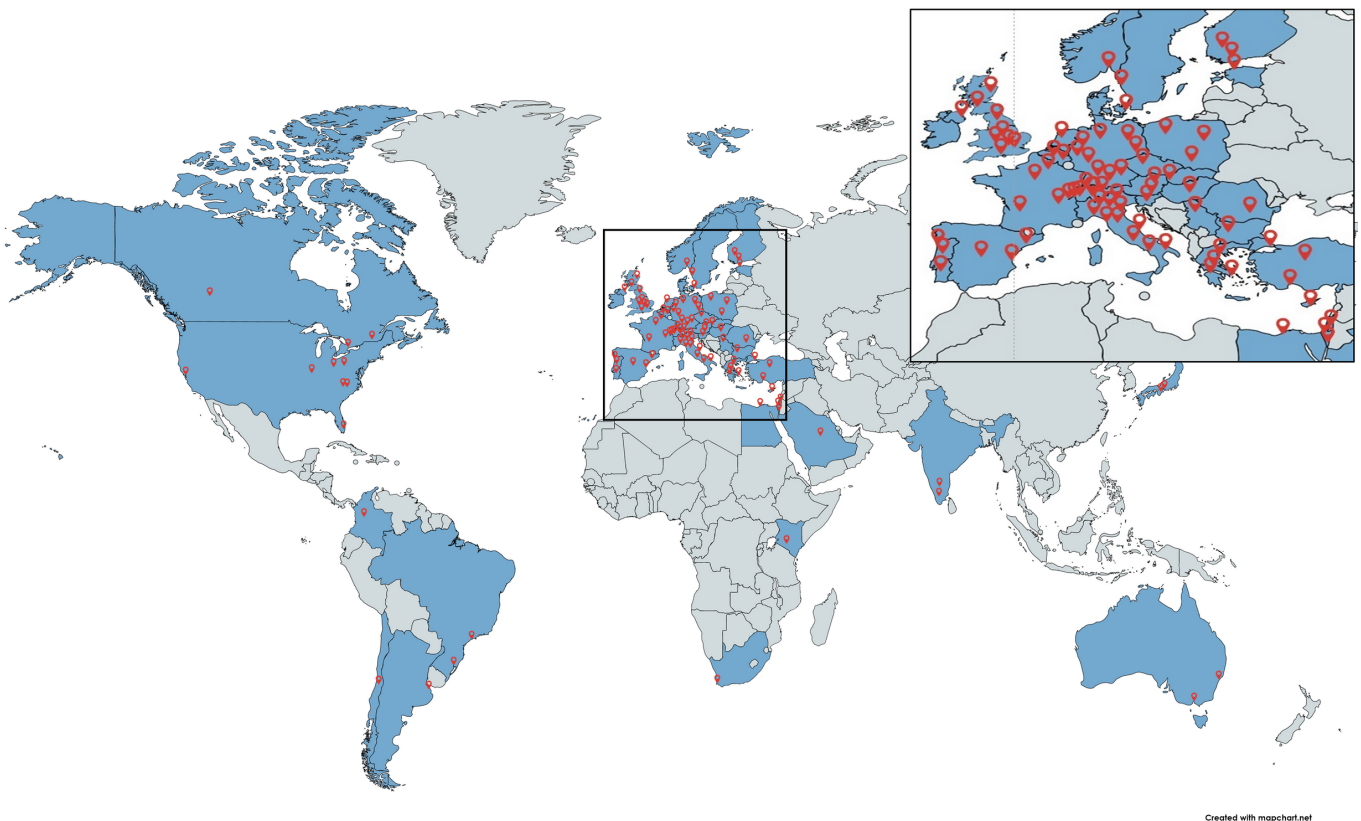
BEAT-PCD Editorial

The BEAT-PCD (Better Experimental Approaches to Treat Primary Ciliary Dyskinesia) Clinical Research Collaboration

Myrofora Goutaki, Suzanne Crowley, Eleonora Dehlink, René Gaupmann, Katie L Horton, Panayiotis Kouis, Yin Ting Lam, Niki T Loges, Jane S Lucas, Jobst F Roehmel, Amelia Shoemark, BEAT-PCD Clinical Research Collaboration

Previous European initiatives, including two European Respiratory Society (ERS) Task Forces (2006-2009/ 2014-2016), an FP7 funded project (BESTCILIA 2012-2015) and the EU COST Action BEAT-PCD (2015-2019) built up and strengthened a multidisciplinary Europe-led network, which includes numerous researchers, clinicians, other health professionals and patient representatives from 27 countries. Despite advances in awareness, diagnosis and clinical care for patients with PCD, diagnosis remains complex with inequalities between countries while treatment is based on expert opinion and is borrowed from other diseases such as cystic fibrosis. Therefore, with this ERS clinical research collaboration (CRC) we aim to advance clinical and translational research in PCD through building upon the foundations set by the previous collaborative initiatives and the BEAT-PCD network. To highlight the aims and objectives of BEAT-PCD we recently published an editorial in the European Respiratory Journal

The **full** text of the editorial is available [here](#).



The BEAT-PCD Clinical Research Collaboration Network. Our network includes >500 individuals from >30 countries (countries shaded in blue and cities marked with red pins in the map) with an interest in PCD.

Early Career Researchers in PCD (ECR-PCD)

Expert Talk series

Apart from the innovative training program for Early Career Researchers offered by BEAT-PCD we also support the ECR network in PCD which has been set-up by the PhD representatives in the BEAT-PCD advisory board. Since March 2021, the ECR network organises a series of Expert Talks by young experts across Europe. These topics range from PCD-specific subjects to skill-building and career advancing lectures.



BEAT-PCD
BETTER EXPERIMENTAL APPROACHES TO TREAT PCD

31st March 2021 at 4PM GMT

Early Career Researchers in PCD Expert Talk

Importance of High-Speed Video Microscopy Analysis in PCD Research

What is this talk about?

The diagnosis of Primary Ciliary dyskinesia (PCD) is challenging because of the absence of a single gold standard test. Both the European Respiratory Society and the American Thoracic Society recognise the importance of High-Speed Video-Microscopy (HSVM) as one of the diagnostic tools. Ciliated cells from nasal or bronchial brushings are recorded through HSVM. Videos can be replayed at slower rate to visualize the cilia motion. Ciliary Beat Frequency (CBF) and/or Ciliary Beat Pattern (CBP) parameters can be calculated.

About Dr Mathieu Bottier:

After a PhD thesis on ciliary beating mechanisms at the University of Paris-Est (France) and a postdoctoral position on Chlamydomonas R. cilia waveform at Washington University in St. Louis (MO, USA), Dr Mathieu Bottier is now a postdoctoral researcher at the University of Dundee (Scotland, UK) working on cilia function in bronchiectasis.

Join us!
Meeting ID: 985 2675 5553
Passcode: 498399
<https://unibe.ch/join/98526755553?pwd=TXA5d1Q5cW56cUJvNFdSemxsenR3dz09>



Dr Mathieu Bottier
Postdoctoral Researcher
University of Dundee

The first Expert Talk was given by Dr Mathieu Bottier, postdoctoral researcher at the University of Dundee, with significant expertise on ciliary beating mechanisms from experiments in human cells and the *Chlamydomonas* animal model. His talk focused on the importance of High Speed Video Microscopy for PCD diagnosis as well as its applications in PCD research. The Expert Talk took place on March 31st 2021.



BEAT-PCD
BETTER EXPERIMENTAL APPROACHES TO TREAT PCD

26th May 2021 at 5PM CET

Early Career Researchers in PCD Expert Talk

"How to apply successfully for an ERS Research Fellowship"

What is this talk about?

The European Respiratory Society (ERS) offers different research fellowships for early career scientists and clinicians. Many of these programmes give an opportunity to develop your own project and visit a research institution abroad to learn new research techniques that are not available at their home institution. Our PCD expert will give an overview of the ERS fellowships programmes focusing on short and long-term fellowships, explain the key elements and give hints about the application process. For more information about key deadlines and information:

<https://www.ersnet.org/education-and-professional-development/funding-opportunities-fellowship-programmes/>

About Dr Myrofora Goutaki:

Dr Goutaki is a medically trained clinical epidemiologist focused on PCD research. She is the co-head of the Swiss PCD registry and leads several PCD projects such as the international ENT PCD cohort study (EPIC-PCD) and she is the co-chair of the BEAT-PCD Clinical Research Collaboration. She has experience with the ERS research fellowship programme as an applicant (short-term ERS fellow) and as a reviewer (member of the ERS college of experts).

Register for the event here!
<https://www.eventbrite.co.uk/e/how-to-apply-successfully-for-an-ers-research-fellowship-by-dr-goutaki-tickets-151072721577>



PD Dr Myrofora Goutaki
Senior researcher
University of Bern

The second Expert Talk was given by Dr Myrofora Goutaki on May 26th 2021 via ZOOM. Dr Goutaki co-chairs the BEAT-PCD ERS CRC and is a medically trained clinical epidemiologist with experience in leading several international PCD projects. Her talk was an overview of ERS short- and long-term fellowship programs and included the key candidate requirements and hints for the application process.

The most recent Expert Talk was given by Ms Holly Keir who is a postdoctoral researcher working at the University of Dundee Ninewells Hospital. She studies neutrophilic inflammation and the lung microbiome in bronchiectasis and COPD patients. Her talk focused on successful science communication to both virtual and in-person audiences. During the same talk an expert poster reviewer (Dr Eric Haarman, University Hospital Amsterdam) provided insights on what is expected by session chairs during poster presentations.



BEAT-PCD
BETTER EXPERIMENTAL APPROACHES TO TREAT PCD

16th June 2021 at 5PM CET

Early Career Researchers in PCD Expert Talk

'A guide to poster preparation and presenting for virtual and in-person conferences'

What is this talk about?

Science communication is key to successful poster presentation and getting the most out of your conference experience. Ms Keir will discuss preparing posters for both virtual and in-person audiences, including tips of presenting to a small group and advice on what chairs look for during a poster session.

Dr Eric Haarman (Paediatric pulmonologist, University hospital Amsterdam) will give insight as an expert poster reviewer.

About Ms Holly Keir

Ms Keir is a postdoctoral researcher working in the Chalmers Lab Group at the University of Dundee Ninewells Hospital, UK. Her research interests are in the role of neutrophilic function and the lung microbiome in patients with bronchiectasis and COPD. She has previously been awarded prizes at the ERS Congress, World Bronchiectasis Conference and British Thoracic Society Meeting for abstract and poster presentations, as well as chairing at a number of poster sessions. For her work on potential treatments for Covid-19 she is nominated for the Postgrad Awards 2021.

Register via Eventbrite:
<https://www.eventbrite.co.uk/e/157586745197>



Ms Holly Keir
Postdoctoral researcher
University of Dundee

BEAT-PCD Website

The BEAT-PCD website is available in English language and is updated regularly. It provides an overview of the network and describes the the project's aims and objectives, introduces the seven Work Packages, provides resources for both health professionals and patients and is frequently updated with BEAT-PCD related research projects, training activities as well as networking and other events.



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Projects

BEAT-PCD supported research projects

For a rare disease such as PCD, international collaboration is essential to improve diagnosis and care. Therefore, BEAT-PCD supports international collaborative projects that aim to fill knowledge gaps and answer important questions for PCD research.

If you would like your project to be supported by BEAT-PCD please contact BEAT-PCD team.

For additional information for each project, check the list below:



Microbial diversity in Primary Ciliary Dyskinesia compared to idiopathic bronchiectasis. A BEAT-PCD and EMBARC biobank study.

Principal Investigator: Stefano Aliberti

Contact Person: Martina Oriano

Quick links

- Resources
- Upcoming Events
- Twitter



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Home

Primary ciliary dyskinesia (PCD) is a rare genetic disease

Cilia that line the mucosal surface are dysfunctional and cannot clear mucus leading to neonatal respiratory distress, persistent daily wet cough, recurrent upper and lower airway infections, bronchiectasis, hearing impairment and persistent rhino-sinusitis.

Cilia are common structures throughout the body, so PCD may affect other organs, for example leading to situs inversus, congenital heart defects and infertility. Mutations in 50 different genes have been identified to date, accounting for approximately 70% of PCD.

News and Quick links

- Short term scientific missions
- Upcoming events
- Working groups

Twitter

In addition, the BEAT-PCD website aims to showcase all PCD related research, published from members of the network. To acknowledge the participation to BEAT-PCD, authors are invited to include the following statement in the declarations section of their manuscript:

"Study authors and data contributors participate in the BEAT-PCD clinical research collaboration, supported by the European Respiratory Society."

Find us online

BEAT-PCD ON SOCIAL MEDIA

