INTRODUCTION

Welcome to the third edition of the BEAT-PCD e-newsletter. In this edition, we highlight PCD-related events at the upcoming ERS International Congress in Milan on 9th-13th September. PCD will be the focus of 5 oral presentations, 16 poster discussions, 3 thematic posters, 1 postgraduate course and 1 industry-sponsored event.

In addition, this e-newsletter highlights 3 recent high-quality publications by BEAT-PCD members, and shines a spotlight on STSMs conducted in the 2015/2016 period (year 1), underlining the long-term impact both in clinical care and research of the training opportunities provided by this important resource.

I look forward to seeing you at the ERS PCD meeting in Milan on 9th September and during the ERS International Congress.

We hope you enjoy this edition of the e-newsletter!

Chair of BEAT-PCD

Jane Lucas

SAVE THE DATE—IMPORTANT PCD EVENTS

- Pre-ERS PCD Meeting, 9th September 2017 from 13:00 to 17:00 at the Aula Magna of Mangiagalli Clinic, University of Milan, Italy
- ERS International Congress, 9th-13th September 2017, Milan, Italy
- 3rd BEAT-PCD Conference & 4th Training School, 6th-9th February 2018, Lisbon, Portugal

PUBLICATIONS FROM BEAT-PCD NETWORK

Please send new publications, achievements, events or activities you would like to see featured on the next e-newsletter edition to b.rubbo@soton.ac.uk. Remember to follow BEAT-PCD on twitter (@beatpcd) and keep up-to-date with the latest developments in the PCD world on the BEAT-PCD official website (http://www.beatpcd.org/).

We look forward to seeing you all in Milan!

BEAT-PCD newsletter team

Bruna Rubbo, Myrona Goutaki, Ana Reula, Panayiotis Kouis, Maciej Dabrowski, Regan Doherty and Florian Halbeisen

12 weeks of inhaled hypertonic saline did not improve St George Respiratory Questionnaire (SGRQ) total score in adult PCD patients in this RCT, but the sample size was small. On the secondary and more disease-specific end-point of the QoL-Bronchiectasis, a significant improvement was observed in the Health Perception scale. This study found little evidence to support the hypothesis that hypertonic saline improves QoL in PCD patients. We advise the use of disease-specific outcome measures in future trials.

http://erj.ersjournals.com/content/49/2/1601770


Immunofluorescence uses antibodies to visualise the presence or absence of ciliary proteins that are often missing in PCD. We found that it is a highly specific diagnostic test for PCD which improves the speed and availability of diagnostic testing, however sensitivity is limited and it is not suitable as a stand.

http://www.atsjournals.org/doi/abs/10.1164/rcpm.201607-1351OC


We have demonstrated the performance of QOL-PCD as a valid and reliable measure for use in clinical trials and clinical practice. The collaboration included researchers, clinicians, and patients from UK, Ireland, USA and Canada. QOL-PCD is the first outcome measure to be validated in PCD, and has already been translated into a number of European and non-European languages.

http://thorax.bmj.com/content/early/2017/02/28/thoraxjnl-2016-209356

**SELECTED PUBLICATIONS**

**CALL FOR APPLICATIONS FOR SHORT-TERM SCIENTIFIC MISSIONS (STSMs)**

The application round for STSMs will be open until January 31st 2018. Fifteen STSMs worth 1,000 Euros each are available for 2017/2018 for training opportunities to both early career researchers and centres looking to strengthen their expertise by bringing over a senior expert to provide in-house training. BEAT-PCD members are encouraged to apply early; please see the application criteria below.

Applications must be made through the COST website: https://e-services.cost.eu/w3/index.php?id=91. Please use the COST Action number BM-1407.

1. Applications must fulfil the following STSM guidelines: “A STSM should specifically contribute to the scientific objectives of the COST Action, while at the same time allowing applicants to learn new techniques or gain access to specific instruments and/or methods not available in their own institutions.”

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3. Supporting documents must include:
   a. A short CV
   b. A supporting letter from your supervisor authorizing the STSM
   c. A supporting letter from the Host Institution, authorizing your trip.

4. Please outline timelines in terms of dates of the visit. STSM’s can be of any duration between 5 and 90 days.

5. All expenses must conform to COST rules. Guidelines all available on the website.

6. Applicants must ensure they have any additional funding needed in place.

7. Applicants must arrange their own health care insurance relevant to the country visited.

8. The STSM is valid only within the financial year of the application. The applicant must have completed the visit and submitted a report of their visit within this financial period.

9. Costs will be reimbursed on receipt of the end of visit report and submission of receipts.

Submissions should be made via email to the Head of the Training School, Claire Hogg (c.hoog@rbht.nhs.uk).

YEAR 1 STSMs (2015-2016): UPDATE AFTER ONE YEAR OF CONCLUSION OF STSM

Sylvia Nyilas from the Bern University Hospital hosted by the University Children’s Hospital Bochum, Germany

STSM: MRI and lung function in patients with PCD

“We performed in thirty-one PCD patients, at the University Children’s Hospital Bochum, a new functional MRI and diverse lung function tests (Nitrogen multiple breath washout and spirometry) on the same day.

We concluded in this project that the combination of functional imaging and lung function tests provides complementary information and are promising measures for lung function impairment in PCD. In a longitudinal setting, we would like to examine the reproducibility and long term variability of the functional MRI and lung function measurements in patients with PCD.

Steps we achieved:
Implementation of SOPs for washout procedures, and training staff for conducting the measurements (by Sylvia Nyilas). Implementation of first functional MRI protocol in Bochum and training MRI staff (by Grzegorz Bauman).

Conducting measurements on PCD patients (by Sylvia Nyilas), adjustment of MRI protocols (by Grzegorz Bauman).

Finalization of measurements on PCD patients, quality control during conduct of measurements by the Bochum staff.”

Oliver Bieri from the University of Basel hosted by the University of Southampton, UK.

STSM: Structural and functional MRI in PCD

Within this STSM joint collaborative lung MRI research projects were initiated between the Division of Radiological Physics at the University Hospital Basel and the PCD Centre at the University of Southampton as well as the Royal Brompton Hospital, and functional MRI
methods have been exchanged and installed at both sites for clinical testing.

Mahmoud Fassad from the University College London hosted by the Institute for Integrative Biology of the Cell, France.

“I spent a month at Dr Anne-Marie Tassin Lab (Gif sur Yvette, France) to study the involvement of novel candidate genes in PCD and cilia motility by analysis of the consequences of gene silencing in a novel PCD model organism, Paramecium. Thanks to the STSM funding scheme that allowed me to pursue such opportunity. I not only was introduced to a new promising PCD model organism that added a lot to my work but I was exposed to a new culture that added a lot to my personal experience as well. “

Panayiotis Kouis from the Cyprus University of Technology hosted by the University of Bern, Switzerland.

“During my STSM at the Institute of Social and Preventive Medicine at the University of Bern (Host Supervisor Prof Claudia Kuehni) I had the opportunity to become involved with the iPCD cohort, a large international cohort study containing baseline and longitudinal data on clinical, diagnostic and treatment for PCD patients. Through the STSM, I gained experience in study protocol development and meta-cohort data management, and I acquired new skills in the field of statistical analysis. Furthermore, we had the opportunity to initiate an international and multicentre study on the frequency determinants and impact of lobectomies in Primary Ciliary Dyskinesia. One year after the initiation of the project, data collection has been completed with a total of 14 PCD centres participating from all around the world and data analysis is currently under way. Preliminary results of the project have been presented during the 2nd BEAT-PCD conference and training school which took place in Valencia in April 2017.”

Amelia Shoemark from the Royal Brompton & Harefield NHS Trust, hosted by the Rikshospitalet in Oslo, Norway.

In the first round of STSMs Amelia Shoemark, a PCD diagnostic scientist at The Royal Brompton Hospital in London, spent five days at Rikshospitalet in Oslo, Norway. The results of this STSM will be presented by Suzanne Crowley, the host, at the European Respiratory Society Conference this year. This STSM also resulted in the initiation of a project for international standardisation of cilia electron microscopy reporting.
ERS PCD meeting Milan 09/09/2017

Time: 13.00-17.00
Location: Aula Magna, Mangiagalli Clinic of the University of Milan.

12:00-13:00  Lunch

13:00  Introduction and greetings  Francesca Santamaria
13:05  BEAT-PCD Action: Update  Jane Lucas
13:15  BEAT-PCD Training school: Update  Claire Hogg

State of the art presentations (30 minutes talk, 10 minutes discussion)

13:25  Advances in chest physiotherapy in primary ciliary dyskinesia: opportunities from cystic fibrosis  Paolo Buonpensiero, Gemma Marsh
14:05  Immunofluorescence analysis use in clinical practice  Amelia Shoemark, Petra Pennekamp

14:45-15:15  Coffee break

Update on collaborative projects (10 min talks, 10 mins discussion)

15:15  PROVALF-PCD study  Bruna Rubbo
15:35  Computer Tomography in adult primary ciliary dyskinesia: typical imaging findings  Jessica Rademacher
15:55  Consensus statement to define pulmonary exacerbations  Jane Lucas
16:15  Consensus statement to standardise testing and reporting of TEM  Amelia Shoemark
16:35  Consensus statement to prevent cross-infections in PCD  Kim Nielsen

17:00  End of the meeting

17:00 onwards  Informal dinner
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<tr>
<th>Day</th>
<th>Location-Session</th>
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<th>Author (country)</th>
<th>8.30</th>
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<tr>
<td>Saturday</td>
<td>Amber 3+4 (South) PG 16 Postgraduate course</td>
<td>Transition in Primary Ciliary Dyskinesia</td>
<td>K. Boisen (Denmark)</td>
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<td>Sunday</td>
<td>Pink (South) Industry sponsored practical workshop: “Hands on FUNC &amp; amp; How to use NO as Biomarker in the Diagnosis and Management of Respiratory Diseases”</td>
<td>Role of nNO to Diagnose PCD</td>
<td>J. Lucas (UK)</td>
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<td>Space 3+4 (South) Paediatric bronchology</td>
<td>PCD Diagnosis by Super resolution (3D SIM) microscopy</td>
<td>S. Dell (Toronto)</td>
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<td>Speed up PCD diagnosis: a quick screening test for recurrent respiratory infections in children</td>
<td>D. Snijders (Italy)</td>
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<td>Primary Ciliary Dyskinesia: first report from Colombia</td>
<td>S. Urcos Rodriguez (Colombia)</td>
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<td>Evolution of Primary Ciliary Dyskinesia (PCD) diagnostic testing in Europe</td>
<td>F. Halbeisen (Switzerland)</td>
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<td>Towards standardised follow-up care for patients with Primary Ciliary Dyskinesia (PCD)</td>
<td>M. Goutali (Switzerland)</td>
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<td>New portable nasal nitric oxide (nNO) analyser differentiates primary ciliary dyskinesia (PCD) from healthy individuals</td>
<td>U. Sepala (Sweden)</td>
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<td>A decision analysis approach for the development of an evidence based diagnostic algorithm for Primary Ciliary Dyskinesia</td>
<td>P. Kouis (Cyprus)</td>
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<td>Ten years’ experience of Primary Ciliary Dyskinesia diagnostic testing</td>
<td>E. Rubbo (UK)</td>
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<td>Monday</td>
<td>Amber 7+8 (South) PCD and air pollution</td>
<td>A high prevalence CDDC103 p.His154Pro mutation causing primary ciliary dyskinesia is associated with normal diagnostic</td>
<td>A. Shoemark (UK)</td>
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<td>Genetic risk factors for laterality defects and congenital heart disease (CHD) in patients with Primary Ciliary Dyskinesia (PCD)</td>
<td>C. Hogg (UK)</td>
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<td>Clinical disease spectrum in RSPH9 Primary Ciliary Dyskinesia patients: a case series</td>
<td>P. Kouis (Cyprus)</td>
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<td>Use of electron tomography to confirm the diagnosis of primary ciliary dyskinesia</td>
<td>A. Shoemark (UK)</td>
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<td>Quality of life in patients with Primary Ciliary Dyskinesia: a systematic review</td>
<td>J. Lucas (UK)</td>
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<td>Spanish version of the Quality of Life Questionnaires for patients with Primary Ciliary Dyskinesia</td>
<td>A. Reula (Spain)</td>
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<td>Bologna (South)</td>
<td>Session ERS Publications: 2017 ERS Guidelines and Task Forces</td>
<td>ERS Guideline for the diagnosis of primary ciliary dyskinesia</td>
<td>J. Lucas (UK)</td>
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<td>TP-20 (Halifax)</td>
<td>Respiratory epidemiology</td>
<td>Diagnosing Primary Ciliary Dyskinesia (PCD) using electron microscopy and exome sequencing</td>
<td>S. Crowley (Canada)</td>
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<td>Blue 2 (North)</td>
<td>Assessing the impact of respiratory and sleep problems in children</td>
<td>Hyperpolarised gas ventilation MRI detects early lung ventilation heterogeneity in children with primary ciliary dyskinesia</td>
<td>L. Smith (UK)</td>
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<td>Functional Magnetic Resonance imaging as a new additional modality in the assessment of Primary Ciliary Dyskinesia</td>
<td>S. Nylas (Switzerland)</td>
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<td>TP-33 (Halifax)</td>
<td>Paediatric broncholeskopy in clinical practice</td>
<td>Primary ciliary dyskinesia and mild cystic fibrosis: lung structure and function similarities</td>
<td>V. Miras (Italy)</td>
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<td>Tuesday</td>
<td>Hypertension in adult patients with primary ciliary dyskinesia</td>
<td>B. Maitre (France)</td>
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<td>Amber 3+4 (South)</td>
<td>Rare and common genetic variants in common and rare airway disease</td>
<td>A rare genetic mutation in primary ciliary dyskinesia</td>
<td>S. Likhova (Russian Federation)</td>
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<td>Preliminary Results of Whole Exome Sequencing in Turkish Primary Ciliary Dyskinesia Patients–Hacettepe University Experience. “Three candidate genes, five novel and two known mutations”</td>
<td>U. Özçak (Turkey)</td>
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<td>Wednesday</td>
<td>Paediatric respiratory infections and immunity</td>
<td>Nasal cavity inflammation in patients with primary ciliary dyskinesia (PCD) is associated with bacterial infection</td>
<td>A. Shoemark (UK)</td>
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<td>Primary ciliary dyskinesia exhibits dysregulated epithelial responses to non-typeable Haemophilus influenzae</td>
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ERS Task Force for Diagnosis of PCD, ERS Congress symposium, 2016