ERS International Congress

On 7th-9th September, our communications officer Katie Dexter attended the virtual ERS International Congress.

What is the ERS International Congress?

The European Respiratory Society Congress is a meeting with approximately 20,000 attendees each year. This conference showcases the excellence across all fields of respiratory medicine. Due to covid-19, this year's congress was held entirely online.

Was PCD discussed?

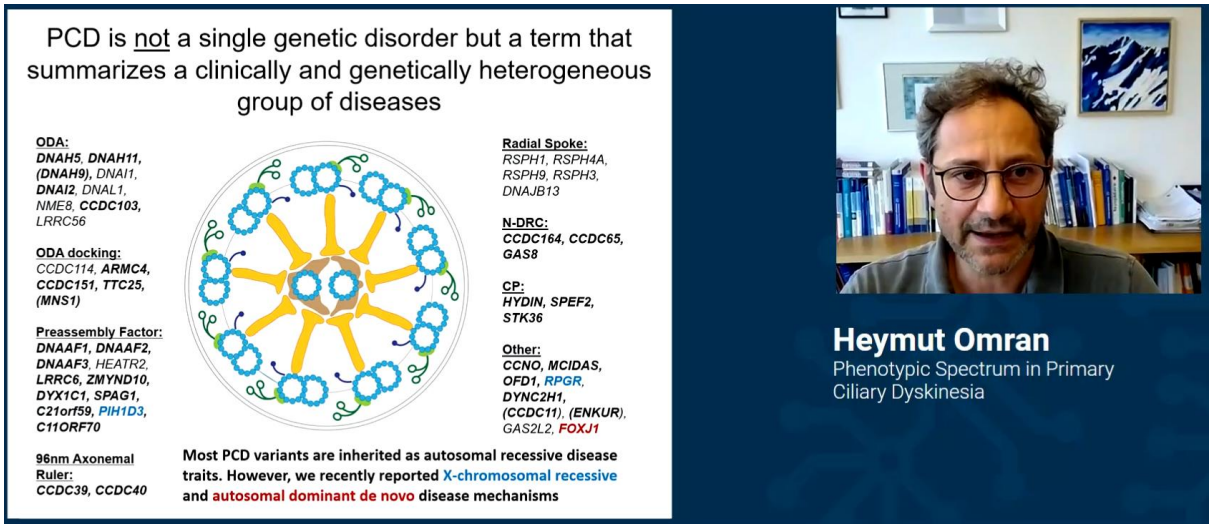
Yes! There were multiple posters and talks on PCD. These ranged from PCD diagnostics to PCD genetics given by researchers as well as clinicians from across the globe. It is humbling to see such a vibrant community interested in our rare disease.

Katie's Highlights

1. PCD Talks and Posters

a) Heymut Omran's overview of PCD

During talks in the European Reference Network for lung diseases (ERN-LUNG) session, Heymut Omran discussed the range of physical structures in PCD, teaching the audience about the many different genetic variations. On top of this, he continued to explain how the diagnosis of PCD is very complex; often requiring multiple different tests. He reiterated that those with normal organ placement are often diagnosed later than those with situs inversus totalis (all organs on the opposite side) or dextrocardia (heart on the right-hand side). Heymut thinks immunofluorescence microscopy (see our latest #PCDLive from Claire and James for an explanation of this!) and genetic testing is the way forward for PCD diagnostics.



b) Playing games to improve lung function and strength

Hazal Sonbahar Ulu presented her work on the "Effects of game-based approach in patients with PCD: a randomized controlled trial." This was an 8-week trial of 32 children, 16 doing active clearance therapy, and 16 to do Active Cycle of Breathing technique as well as 40 minutes of Xbox Kinect 360 games three times a week. Hazal shows that the group with games have improved lung function, respiratory muscle strength, and exercise capacity. Hazal says that this is likely due to this exercise being more preferable as it is more fun. We all know that exercise is vital for your lung health so remember to keep fit in whatever manner suits you!

c) Genetic testing for Diagnosing PCD

Sandra Rovira Amigo (Barcelona, Spain) spoke about the "Implementation of a gene panel for the genetic diagnosis of PCD."

In this study, 79 patients were included with 53 of those diagnosed with confirmed or highly likely PCD. The most common causal genes were DNAH5 and CCDC39, in patients of Caucasian origin, while in non-Caucasian ones the most common genes were CCDC40, DNAI2 and RSPH4A. They found 52 different gene variants which caused PCD, 36 not previously described in the literature. Sandra says that the design and implementation of a specific gene panel (i.e. looking at the particular genes involved in PCD rather than all of the genes) has a high efficiency for the genetic diagnosis of PCD.

d) PCD Posters!

On top of the many amazing talks, there were a large number of posters dedicated to PCD. Posters are usually hung up on walls with the author standing beside to chat informally about their work. This year, due to the virtual style, author's have pre-recorded a short 2-3-minute explanation and viewers are able to submit questions and comments about their poster online for all to see and discuss. A few posters of the posters are summarised below.

Jane Lucas (Southampton) explained the results of the Wessex PCD cohort of the 100,000-genome project where 21 PCD patients, and 55 patients with non-CF bronchiectasis were investigated. 70% of patients with no definitive PCD diagnosis were confirmed as PCD in this study, as well as 8% of the adult non-CF bronchiectasis diagnosis were actually found to have PCD.

Amanda Harris (Southampton) explains the experience of using nasal nitric oxide (nNO) testing in a national PCD centre where she explains that nNO is a good screening tool for PCD but should not be used alone for diagnostics. She explains that more work on technical standards are required to develop this tool further in PCD diagnostics.

New genetic markers are being discovered in Turkey; Ayse Tana Aslan describes a novel mutation in PCD discovered in two siblings. Work being done at the Royal Brompton Hospital by Andreia Lucia do Nascimento Pinto hopes to help diagnose PCD patients by using artificial intelligence to assist with TEM analysis. Marieke Varkleij explains psychological support is needed in PCD after assessing anxiety and depression in 72 Dutch PCD patients.

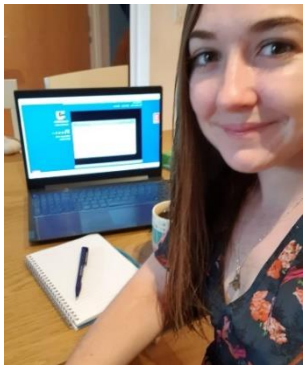
2. Patient involvement

Patients were involved throughout the conference and often short videos of patient experiences were played at the start of the sessions. Our PCD volunteer and committee member Susi Shanks presented at the patient networking day and helped explain to other patient groups how the PCD Family Support Group has adapted to the challenges of covid-19

Ron Flewett (59)**:** Diagnosed at 53, tells us how afraid and frightened he was during diagnosis and how important it was to have other people to talk to with similar experiences. A quote we can all relate to: "sometimes when I wake up, I don't feel like exercising […] but when I'm finished it makes me feel good and lifts my mood."

Read more about Ron: https://twitter.com/EuropeanLung/status/1303311990639730688

About the author



Katie Dexter is 29 and has PCD. She is a scientist, graduating with a PhD in Physics from the University of Leicester in 2018. Her job is in preclinical imaging where she works with MRI, CT, and nuclear medicine techniques in cancer research. Katie attended this conference as a volunteer, on behalf of the PCD Family Support Group UK. Pictured is Katie in her home in Leicester attending the conference remotely.

Get in touch

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