Living with PCD: By and for people with PCD

Updates and plans

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Living with PCD

Together with Primary Ciliary Dyskinesia (PCD) support groups from all over the world we set up the *Living with PCD* (previously COVID-PCD) study in 2020.

The *Living with PCD* study collects data through online anonymous questionnaires. So far, we studied COVID-19 in people with PCD, vaccinations against COVID-19, PCD diagnostic tests, fertility, fertility care, physical activity, and flare-ups of lung symptoms in people with PCD. Participants receive a detailed questionnaire when they start the study and extra questionnaires on special topics from time to time.

Join *Living with PCD*: Anyone with PCD from anywhere in the world who did not yet participate can join the study via pcd.ispm.ch

Partners

University of Bern, Switzerland; University of Southampton, UK; Selbsthilfegruppe Kartagener Syndrom und Primäre Ciliäre Dyskinesie, Switzerland; PCD Support UK; Verein Kartagener Syndrom und PCD, Germany; PCD Australia; PCD Foundation, USA; Asociación Española de Pacientes con Discinesia Ciliar Primaria, Spain; Associazione A.I.D. Kartagener Onlus, Italy; and Association ADCP, France.

Study Funding from

- Swiss National Science Foundation (192804, 10001934)
- Swiss Lung Association
- PCD Foundation, USA
- Verein Kartagener Syndrom und Primäre Ciliäre Dyskinesie, Germany
- PCD Support UK
- PCD Australia

Living with PCD: Building momentum for exciting times ahead

Thank you for taking part in this study. We greatly appreciate the time and effort you have put into completing questionnaires and hope you will continue to participate in the future.

While we did not send out any new questionnaires last year, it was by no means a quiet year for the *Living with PCD* study. We focused on preparing for the future, analysed existing data (see page 2), published findings in scientific journals (page 3) and presented our work at various conferences (page 3).

As part of the transition from COVID-PCD to *Living with PCD*, we're updating our study procedures and getting new ethical approval. We'll update the website once everything is finalised—thank you for your patience!

We are excited to announce funding from the Swiss National Science Foundation, awarded to Myrona Goutaki, which will enable us to continue the study and expand our team (page 4).

We are also pleased to introduce you to some of the *Living with PCD* team members in this newsletter so that you get to know us better (page 4).

As always, we welcome your questions, feedback, and suggestions for the study. If you have ideas for a new questionnaire or other input, please feel free to email us at pcd.ispm@unibe.ch. We look forward to hearing from you!

With thanks,

The *Living with PCD* study team from the Institute of Social and Preventive Medicine at the University of Bern in Switzerland: (from left) **Myrona Goutaki** (research team leader, senior researcher), **Leonie Schreck** (PhD student), **Claudia Kuehni** (group leader, professor), **Andrea Fernandez** (PhD student).

Results - what research questions did we address during the past year?

Last year, we focused on the relationship between PCD genes, symptoms and flare-ups of lung symptoms. We asked the following questions:

- How do the PCD genes of our participants relate to their symptoms?
- How often do people with PCD have flare-ups of lung symptoms, and which individuals tend to have more?

How do PCD genes relate to our participants' symptoms?

Errors in many different genes can cause PCD. It is important to understand which PCD genes relate to which symptoms. This would allow people to receive a different appropriate treatment depending on their genetic PCD type. We analysed data from 206 participants who knew the gene that causes their PCD. The most common genes were DNAH5 (34 %), DNAH11 (13 %), CCDC40 (10 %), DNAI1 (9 %), CCDC39 (6 %) and RSPH1 (4 %). All participants had symptoms such as cough, frequent lung infections, ear pain, and shortness of breath. However, the age at diagnosis and if they had their organs mirrored (called situs inversus) varied by gene.

Read more about these results here.

How often do people with PCD have flare-ups of lung symptoms, and which individuals tend to have more?

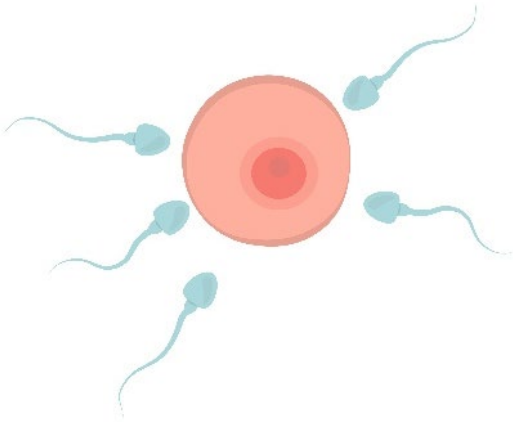
Flare-ups of lung symptoms, often due to chest infections, are common in people with PCD. However, it is unclear how often people with PCD have flare-ups. We also do not know which individuals tend to have more. In the *Living with PCD* study, 660 participants, including adults, teens, and children, reported their symptoms every week for two years. We found that people with PCD had on average three flare-ups per year. The frequency was similar for adults and children. However, adult women and people with the bacterium *Pseudomonas aeruginosa* found in their sputum had more flare-ups. Half of the participants managed their flare-ups at home without contacting a health professional. They adjusted their treatments, such as starting antibiotics or doing more physiotherapy. These findings show how much flare-ups affect people with PCD and emphasise who might need more personalised care.

Read more about these results here.

Publications – what was published?

We are very pleased to announce that the *Living with PCD* team has already published nine articles in scientific journals. You can read all our short summaries in everyday language and the full articles in scientific English on our website: pcd.ispm.ch/en/publications/

Are you interested in what we found out about **fertility**?



Our latest publication answers the question "How do people with PCD and their caregivers report their experiences with fertility?"
Read the summary here:

"Nobody has ever spoken to me about PCD and fertility issues": Fertility experiences of people with primary ciliary dyskinesia and their caregivers

Read more about our other publications on fertility in people with PCD:

Infertility and pregnancy outcomes among adults with primary ciliary dyskinesia

Fertility care among people with primary ciliary dyskinesia

Communication - How do we share our results?

During the past year, we presented results at congresses and meetings. In Vienna, we participated in the PCD Annual Research Meeting (https://beat-pcd.squarespace.com/events) and the European Respiratory Society congress (https://www.ersnet.org/congress-and-events/congress/) where we presented results on fertility and flare-ups of lung symptoms to health professionals and researchers. We presented data on fertility also at the 2024 PCD On The Move Conference in Puerto Rico and at the Swiss Pulmonology Conference to lung doctors.

At these events, we had the opportunity to reach and engage with a wide audience. We gave talks to an audience of several hundred, participated in panel discussions and had one-on-one conversations with experts and patients. We also presented posters summarizing our research findings, which gave us the opportunity to discuss our results in an interactive format with interested colleagues. This exchange was invaluable as it helped us to disseminate our work and gather new perspectives to improve future research.

Study team – how did we start with PCD research?

In this section, two members of our study team introduce themselves, describe their research focus, and their experiences working on the *Living with PCD* study.

Claudia Kuehni

"How I Became a PCD Researcher: I'm Claudia Kuehni. I live in Switzerland, and I started as a general paediatrician like my father. Later I specialised in respiratory diseases. My PCD adventures began by chance in 2006 when I was asked to provide statistical expertise for the 1st European Respiratory Society's PCD task force. This task force sparked an avalanche of studies, transforming PCD research from a little-known area into a fast growing field.

Our *Living with PCD* study also started spontaneously. At the pandemic's start in 2020, Bernhard Rindlisbacher, a dear friend, called to share PCD patients' concerns about COVID-19 and suggested studying its effects. Together with him, with Eva Pedersen, and with many others, we designed the study in record time over the Easter holidays, launching it within months. Thanks to all of you, this participatory study is an incredible experience—and I hope it continues!

"I am Leonie. I am a medical doctor currently doing a PhD on PCD at the University of Bern. I have been managing the *Living with PCD* study for the past three years. I have chosen to do research on PCD because I want to improve care for people with rare diseases. I will complete my doctorate at the end of February 2025, which also means the end of my work on the *Living with PCD* study. After that, I plan to complete my specialization in public health in Switzerland and hope to return to rare disease research in the future. The insights and encouragement I have received from the patient advisory group and the study participants have been invaluable and an important source of motivation and learning throughout my doctoral studies."

Leonie Schreck

Future plans of the *Living with PCD* study

Myrona Goutaki

Myrona Goutaki has been awarded a grant from the Swiss National Science Foundation (https://data.snf.ch/grants/grant/10001934), which will allow to design a series of questionnaires to explore the social and psychological well-being of people with PCD and their families and identify the key factors that influence their quality of life. To do this, she has recruited **Andrea Fernandez**, a new PhD student, and **Nena Karavasiloglou**, a postdoctoral fellow.

Participation

How can you be involved in the *Living with PCD* study?

We appreciate your participation in the *Living with PCD* study, and we hope you want to continue. If you would like to influence and shape the *Living with PCD* study, this is possible in several ways:

- Send us your feedback about how it is to participate in the study – what can we improve?
- Send any suggestions you have for new questionnaires. We especially welcome suggestions related to the topic of social and psychological well-being of people with PCD, which will be the focus of the next questionnaires. We want to ensure to capture your ideas and feedback.
- Tell us if you want to participate in an advisory group that meets twice to three times yearly to discuss new ideas.

You are also welcome to ask any questions you may have about the study. Our email address is: pcd.ispm@unibe.ch.

We look forward to hearing from you!

Partners:

Who are the partner institutions and groups for the *Living with PCD* study?

Australia: PCD Australia

France: Association ADCP

Germany: Verein Kartagener Syndrom und PCD

Italy: Associazione A.I.D Kartagener Onlus

Spain: Asociación Española de Pacientes con Discinesia Ciliar Primaria

Switzerland: Selbsthilfegruppe Kartagener Syndrom und PCD

United Kingdom: PCD Support UK

USA: PCD Foundation

Switzerland: University of Bern

United Kingdom: University of Southampton

International PCD network: BEAT-PCD

Curious to learn more about PCD research?

If you're interested in exploring ongoing research on PCD beyond the *Living with PCD* study, we invite you to visit the BEAT-PCD website at: https://beat-pcd.squarespace.com/. There, you'll find valuable insights into additional research initiatives and collaborations in the PCD community.

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