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# Genotypes and associations with symptoms in primary ciliary dyskinesia

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**Why did we do this research and why is it important?**

It is important to understand the role of specific genetic mutations (changes in the DNA) in primary ciliary dyskinesia (PCD). If we better understand which mutations lead to which symptoms, people can get access to the appropriate treatment depending on their genetic type of PCD. We studied how the more than 50 different genes that cause PCD relate to self-reported symptoms and characteristics such as age of PCD diagnosis and laterality defects (one or more organs situated in the wrong side of the body).

**How did we do this research?**

We used data from the COVID-PCD study, an online study collecting information directly from people with PCD from all over the world. People with PCD registered online to the study and then received a questionnaire via email. We asked questions about PCD genes, symptoms, and medical history.

**What did we find out?**

Of the 759 people in the study, 444 had genetic testing, and 289 wrote that a PCD gene was found. We focused on 206 people who knew which gene was causing their PCD. The most common genes causing PCD were DNAH5 reported by 34% of the 206 people, DNAH11 (13%), CCDC40 (10%), DNAI1 (9%), CCDC39 (6%), and RSPH1 (4%). We grouped these genes based on what part of the cilia (tiny hair-like structures in our body) they affect. The biggest group was the one affecting the dynein structure which helps the cilia move (reported by 62%). We found that the age of PCD diagnosis and frequency of laterality defect varied between groups. However, most people reported experiencing symptoms daily or often, regardless of PCD gene group.

**What does it mean?**

Our study showed that different genes causing PCD may lead to specific health problems, but symptoms like cough, ear pain, and shortness of breath are common among all people with PCD, regardless of the gene.

**Further information: pcd.ispm.ch**

**Full article reference:** Pedersen ESL, Goutaki M, Schreck LD, Rindlisbacher B, Dixon L, Lucas JS, Kuehni CE; COVID-PCD patient advisory group. Questionnaire-assessed genotypes and associations with symptoms in primary ciliary dyskinesia. ERJ Open Res. 2024 Oct 28;10(5):00288-2024.
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You can find the full article in English here.

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COVID-PCD