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Diagnostic testing in people with primary ciliary dyskinesia: an international study

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

Primary ciliary dyskinesia (PCD) can be diagnosed with different tests, such as measuring a special gas in the nose, the so-called nasal nitric oxide (nNO) test, examining cells taken from inside the nose under the microscope (biopsy), or genetic tests looking for a defect that causes PCD. This diagnosis usually requires several tests, but in some individuals the diagnosis can be confirmed by genetic testing or biopsy. However, a combination of tests is necessary to define different types of PCD, which may require different treatment plans and monitoring. This knowledge is also important for developing personalized treatments and participating in clinical trials. We wanted to find out which diagnostic tests people with PCD have had, and possible explanations why some people have tests done and others not.

How did we do this research?

We used data from COVID-PCD, an international study for and by people with PCD. People with PCD registered online to the study and then received a questionnaire via email. The questionnaire asked about diagnostic tests and about other factors such as the year of their PCD diagnosis and if they have mirrored organs (situs inversus).

What did we find out?

Among 747 people with PCD from 49 countries, half (49%) had nNO measured, three-quarters (75%) had biopsy samples taken from their nose or lungs, and more than half (58%) had done a genetic test. This differed between countries: In Switzerland only one-third had a genetic test done but in North America, two-thirds had. One-third (36%) of all participants had all three tests done. Participants diagnosed with PCD a long time ago (before the year 2000) and participants with mirrored organs had less tests done than others.

What does it mean?

Not all people with PCD in the world have the same diagnostic tests done. For many people, PCD diagnosis is based only on a single test or even only on clinical features such as mirrored organs. These people could benefit from additional tests so that we better understand their type of PCD, and they can profit from more personalized treatments in the future.

Further information: www.covid19pcd.ispm.ch

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You can find the full article in English here.

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COVID-PCD