

## Asthma more prevalent among children with a rare genetic disorder, study finds



**Researchers said understanding the overlap between primary ciliary dyskinesia and asthma is critical to helping young patients have healthier respiratory outcomes. Credit: Indiana University**

Indiana University (IU) School of Medicine researchers have uncovered compelling evidence that children with primary ciliary dyskinesia (PCD), a rare genetic disorder affecting airway function, are significantly more likely than children without PCD to have asthma.

The findings, recently [published in a JAMA Network Open research letter](#), highlight the importance of routine asthma screening for children with PCD and suggest some children with asthma may have undiagnosed PCD.

An estimated 1 in 10,000 to 30,000 people in the United States have PCD. It affects the microscopic, hair-like structures called cilia that line the airways and help clear mucus, leading to an increased risk of serious breathing problems and infections in people with the inherited condition.

"The connection between PCD and asthma has not previously received much attention," said Benjamin Gaston, MD, the Billie Lou Wood Professor of Pediatrics at the IU School of Medicine, who co-led the study. "Our data analysis revealed an undeniable link, showing children with PCD were 22 times more likely to have asthma compared to children without PCD characteristics."

In addition to Gaston, the study team includes co-leader Arthur Owora, Ph.D., MPH, associate professor of pediatrics at the IU School of Medicine and a research scientist at the Regenstrief Institute, along with collaborators from the Mayo Clinic and Case Western Reserve University.

The team analyzed 266 pediatric participant records from two large electronic health record databases: the Indiana Network for Patient Care Research and TriNetX. They compared a non-PCD control group against children with bronchiectasis and situs inversus totalis, two hallmark conditions strongly associated with PCD.

Across both data cohorts, asthma was significantly more prevalent among children with PCD. The findings also suggest that some children diagnosed with asthma may have undiagnosed PCD, particularly if they exhibit unique rhinitis symptoms or persistent respiratory issues.

"Understanding the overlap between PCD and asthma is critical to helping young patients have healthier respiratory outcomes," said Gaston, who is also a pediatric pulmonologist at Riley Children's Health. "Future research, including clinical observations and formal airway reactivity tests, will be essential to further explore this relationship."

**More information:** Joe Zein et al, Asthma Among Children With Primary Ciliary Dyskinesia, *JAMA Network Open* (2024). DOI: [10.1001/jamanetworkopen.2024.49795](https://doi.org/10.1001/jamanetworkopen.2024.49795)

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