European Respiratory Society and American Thoracic Society guidelines for the diagnosis of primary ciliary dyskinesia.

Guideline ID: 62 Published: 2025 Dec 18

European Respiratory Society (ERS) | American Thoracic Society (ATS)

Shoemark A, Goutaki M, Kinghorn B, et al. European Respiratory Society and American Thoracic Society guidelines for the diagnosis of primary ciliary dyskinesia. Eur Respir J. 2025 Dec 18;66(6):2500745. [80 references] PubMed

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Overview

Guideline Objective

To provide a unified, updated joint ERS/ATS clinical practice guideline regarding how to diagnose or refute the diagnosis ofprimary ciliary dyskinesia (PCD)

Patient Population

Patients suspected to have PCD

Recommendations

Recommendation Statements

Population, Intervention, Comparison, and Outcome (PICO) 1: Should patients suspected for primary ciliary dyskinesia (PCD) be tested with nasal nitric oxide (nNO) in addition to transmission electron microscopy (TEM) and/or genetics?

PICO 1a: Should patients suspected for PCD be tested with nNO measurement during velum closure (breath hold; exhaling against resistance) in addition to TEM and/or genetics?

The panel recommends the use of measurement of nNO (during velum closure) in addition to genetics and/or TEM for the diagnosis of PCD among patients suspected to have PCD. (**Strong recommendation for the intervention; Moderate certainty of evidence**)

PICO 1b: Should patients suspected for PCD be tested with nNO measurement during tidal breathing in addition to TEM and/or genetics?

The panel suggests the use of measurement of nNO (during tidal breathing) in addition to genetics and/or TEM for the diagnosis of PCD among patients suspected to have PCD. (**Conditional recommendation for the intervention; Very low certainty of evidence**)

PICO 2: Should patients suspected for PCD be tested with ciliary beat pattern assessment using high-speed video microscopy (HSVM) in addition to TEM and/or genetics?

PICO 2a: Should patients suspected for PCD be tested with ciliary beat pattern assessment of post-cell culture using HSVM in addition to TEM and/or genetics?

PICO 2b: Should patients suspected for PCD be tested with ciliary beat pattern assessment of pre-cell culture using HSVM in addition to TEM and/or genetics?

The panel recommends ciliary beat pattern assessment using HSVM in addition to TEM and/or genetic testing to diagnose PCD. (**Strong recommendation for the intervention; Very low certainty of evidence**)

Although the analyses for PICO2a and PICO2b were done separately, the panel decided on a joint recommendation.

PICO 3: Should patients suspected for PCD be tested with immunofluorescence (IF) in addition to TEM and/or genetics?

The panel recommends IF in addition to TEM and/or genetic testing to diagnose PCD. (**Strong recommendation for the intervention; High certainty of evidence**)

Narrative 1: What are the clinical manifestations in the newborn period, childhood and adulthood driving a decision to refer a patient for PCD diagnostic testing?

Commonly reported characteristics include neonatal respiratory distress in a term infant, a persistent wet cough from early childhood, year-round rhinosinusitis, serous otitis media and hearing loss, bronchiectasis, infertility/subfertility and laterality/heterotaxy defects. A systematic review and meta-analysis confirmed that clinical symptoms of individuals with PCD are highly variable, reflecting true heterogenicity as well as methodological biases.

Narrative 2: What additional diagnostic tests could be useful for diagnosing PCD?

Additional diagnostic tools can be considered to further improve diagnostic accuracy in PCD, and these tests can be broadly categorised into genetic and clinical tests.

Narrative 3: Overcoming PCD diagnostic challenges in resource-limited settings: what strategies work?

Given the complexity and multifaceted approach to PCD diagnosis, resource-limited settings present additional challenges and obstacles in making the diagnosis, because expertise in PCD and specific PCD diagnostic tests vary depending on country and region. We defined resource-limited settings as those characterised by a lack of funds to cover healthcare costs, on an individual or societal basis, leading to limited access to

medication, equipment, supplies, devices and/or expertise for specific diseases (i.e., PCD).

Strategies to circumvent barriers to PCD diagnosis in resource-limited settings include collaboration with established PCD centres for procedural training (nasal ciliary biopsy, nNO testing), diagnostic expertise (TEM, HSVM and IF) and access to funding mechanisms for expanded genetic testing. Additionally, networks such as the BEAT-PCD, ERN-LUNG and the Genetic Disorders of Mucociliary Clearance Consortium, and patient foundations including the North American PCD Foundation and PCD Support UK, can help with the diagnosis and management of PCD by supporting local physicians. There is a knowledge gap for PCD testing in resource-limited areas; epidemiological studies/surveys are needed to evaluate the barriers and knowledge gaps among physicians in low-resource countries, and existing tools and innovative diagnostic solutions need validation.

Evidence Rating Scheme

Quality of Evidence Grades and Their Definitions*

Quality	Definition
High	We are very confident that the true effect lies close to that of the estimate of the effect.
Moderate	We are moderately confident in the effect estimate: The true effect is likely to be close to the estimate of the effect, but there is a possibility that it is substantially different.
Low	Our confidence in the effect estimate is limited: The true effect may be substantially different from the estimate of the effect.
Very Low	We have very little confidence in the effect estimate: The true effect is likely to be substantially different from the estimate of effect.

* Schünemann H, Brożek J, Guyatt G, Oxman A, editors. GRADE Handbook for Grading Quality of Evidence and Strength of Recommendations. Updated October 2013. The GRADE Working Group, 2013.

Recommendation Rating Scheme

Strength of Recommendations and Their Rationale[1,2]

Recommendation Strength	Rationale
Strong recommendation for	The panel is certain that desirable outweigh the undesirable effects
Conditional recommendation for	The panel is less confident that desirable outweigh the undesirable effects
Conditional recommendation against	The panel is less confident that undesirable outweigh the desirable effects
Strong recommendation against	The panel is certain that undesirable outweigh the desirable effects
No recommendation	The confidence in the results might be very low to make a recommendation, or the trade-offs between the desirable and undesirable effects are finely balanced. Or no data is available.

1 Andrews J, Guyatt G, Oxman AD, Alderson P, Dahm P, Falck-Ytter Y, et al. GRADE guidelines: 14. Going from evidence to recommendations: the significance and presentation of recommendations. J Clin Epidemiol. 2013;66(7):719-25.

2 Schünemann H, Brożek J, Guyatt G, Oxman A, editors. GRADE handbook for grading quality of evidence and strength of recommendations. Updated October 2013. The GRADE Working Group, 2013.

Related Content

Supporting Documents

- Supplementary Material.
- Using the GRADE Approach for Guideline Development: A Step-by-Step Guide.

- ERS Handbook for Clinical Practice Guidelines: Methodological Guidance for Developing ERS Clinical Practice Guidelines; 2021 Nov 11.
- ERS Task Forces for the Development of Clinical Practice Guidelines, Statements and Technical Standards – Guidance 2025-2026; 2025 Nov.

Implementation Tools

- One-Page Guideline; 2025.
- Pocket Guideline; 2025.
- The Original Guideline includes the following:
    - Figure 4. Graphical Presentation of Proposed Algorithm for the Diagnosis of Primary Ciliary Dyskinesia (PCD)

Patient Education

No patient education materials available.

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TRUST Scorecard

Composition of Guideline Development Group (GDG)

Multidisciplinary GDG Members	**Yes**
Methodologist Involvement	**Yes**
Incorporation of Patient and Public Perspective	★★★★★

Systematic Review of Evidence

Literature Search	★★★★★
Study Selection	★★★★★
Evidence Synthesis	★★★★★

Foundations for Recommendations

Strength of Evidence Grade	★★★★★
Description of Benefits and Harms of Recommendations	★★★★★
Summary of Evidence Supporting Recommendations	★★★★☆
Strength of Recommendations Rating	★★★★☆
Clear Articulation of Recommendations	★★★★★

Funding Source — **Yes**

Disclosure and Management of Financial Conflicts of Interests — ★★★★★

External Review — ★★☆☆☆

Updating — ★☆☆☆☆

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