

## Peripheral Microtubule Doublets Tilt: Questioning Normal Ciliary Ultrastructure in Primary Ciliary Dyskinesia (PCD) Patients

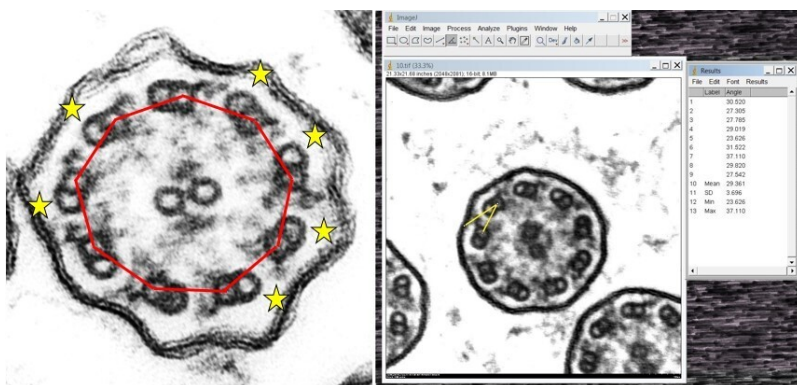
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### Abstract

Assessment of ciliary ultrastructural defects by TEM plays a key role in the diagnostic procedure for PCD. However, about 30% of patients exhibit normal ciliary ultrastructure (NCU), and this proportion is increasing with the current molecular advances. During our routine analysis of ciliary axonemes, we detected and registered in all PCD patients a visible tilting of more than 3 peripheral microtubule doublets (PMD) in many of the analyzed ciliary sections (figure 1). Given the subjective interpretation of this finding, we considered quantitatively determining the PMD deviation by assessing its tilting angle (figure 1). In this study, we aimed to investigate this ultrastructural feature in NCU PCD patients' cilia, testing the hypothesis that NCU patients show deviation in most of their doublets.

For that purpose, we evaluated the tilt angle of the 9 PMD in 10 ciliary sections per participant in nasal brushings of NCU patients with confirmed PCD (n=15) compared to healthy controls (n=15). The mean PMD angle in healthy ciliary sections was about 25.5 degrees, and the number of tilted PMD above this value in NCU PCD patients was significantly higher than in controls (p<0.001). If confirmed, this technique could provide a novel diagnostic approach for PCD.



Micrograph showing peripheral microtubule deviation (left). Microtubule angle quantification using ImageJ (right).

### References

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