

# Repeating ciliary videomicroscopy improves the specificity for PCD diagnosis

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**Background:** Primary ciliary dyskinesia(PCD) is an inherited motor ciliopathy in which respiratory cilia are dyskinetic. High speed videomicroscopy analysis (HSVA) allows to evaluate beat frequency(CBF) and pattern(CBP). HSVA is highly sensitive and specific for PCD when combining CBF and CBP evaluation, as CBF alone lacks sensitivity and specificity. Currently, only electron microscopy(TEM) and genetics are recognized as confirmatory diagnostic tests for PCD. ERS guidelines state that HSVA should be repeated on 3 separate occasions to suggest a PCD diagnosis. However, the 3 visits imposed on the patient constitute a heavy burden and a significant cost.

**Aims:** To compare the sensitivity and specificity for PCD diagnosis of 1 HSVA evaluation versus HSVA repeated on 3 separate occasions.

**Methods:** We defined PCD positive if TEM and/or genetics were positive, and PCD negative if TEM and genetics were negative. We selected patients who had successful HSVA on 3 separate occasions. HSVA was considered as abnormal if the percentage of abnormal CBP was higher than our laboratory normal values. We compared the sensitivity and specificity between the first HSVA and the 3 repeated HSVA. We considered that the results of 3 HSVA was positive if the patient had 3 abnormal HSVA, and negative if the patient had  $\geq 1$  normal HSVA.

**Results:** 10 patients(4 PCD positive and 6 PCD negative)had 3 successful HSVA. The sensitivity of 1 HSVA versus 3 repeated HSVA to diagnose PCD were similar (75%). However, the specificity of 3 repeated HSVA was higher than a single HSVA to diagnose PCD (75% vs 33%, respectively).

**Conclusion:** This pilot study suggests that repeating HSVA on 3 separate occasions improves the specificity of the test.