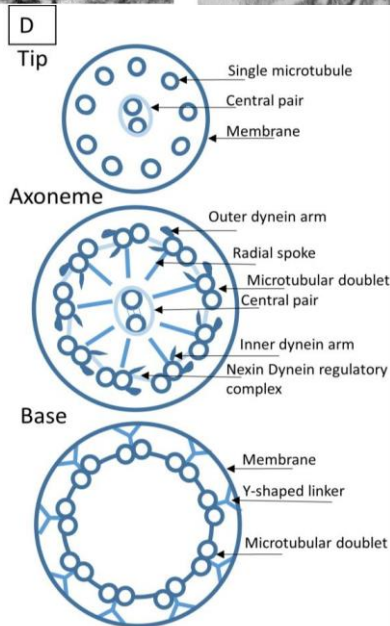
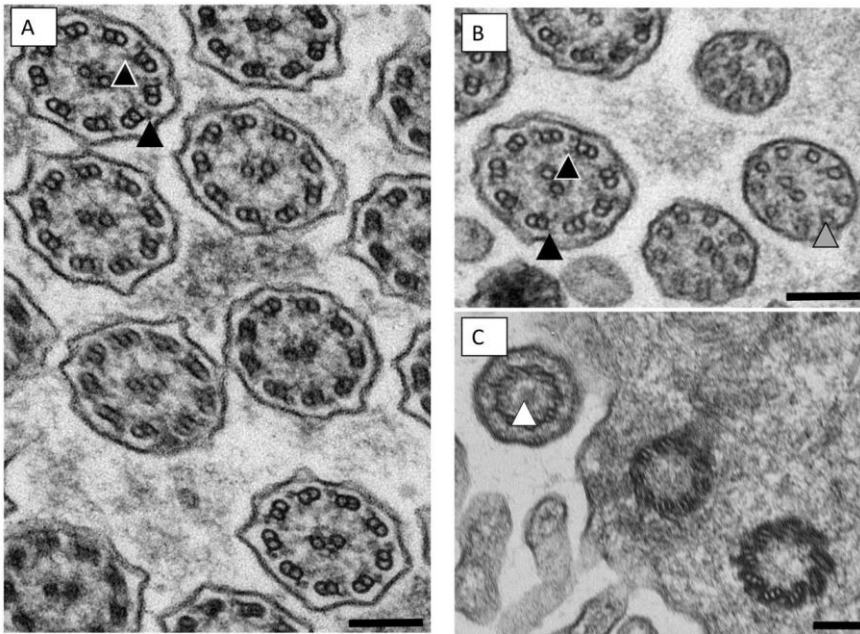


A quick guide to:

# Transmission Electron Microscopy guidelines

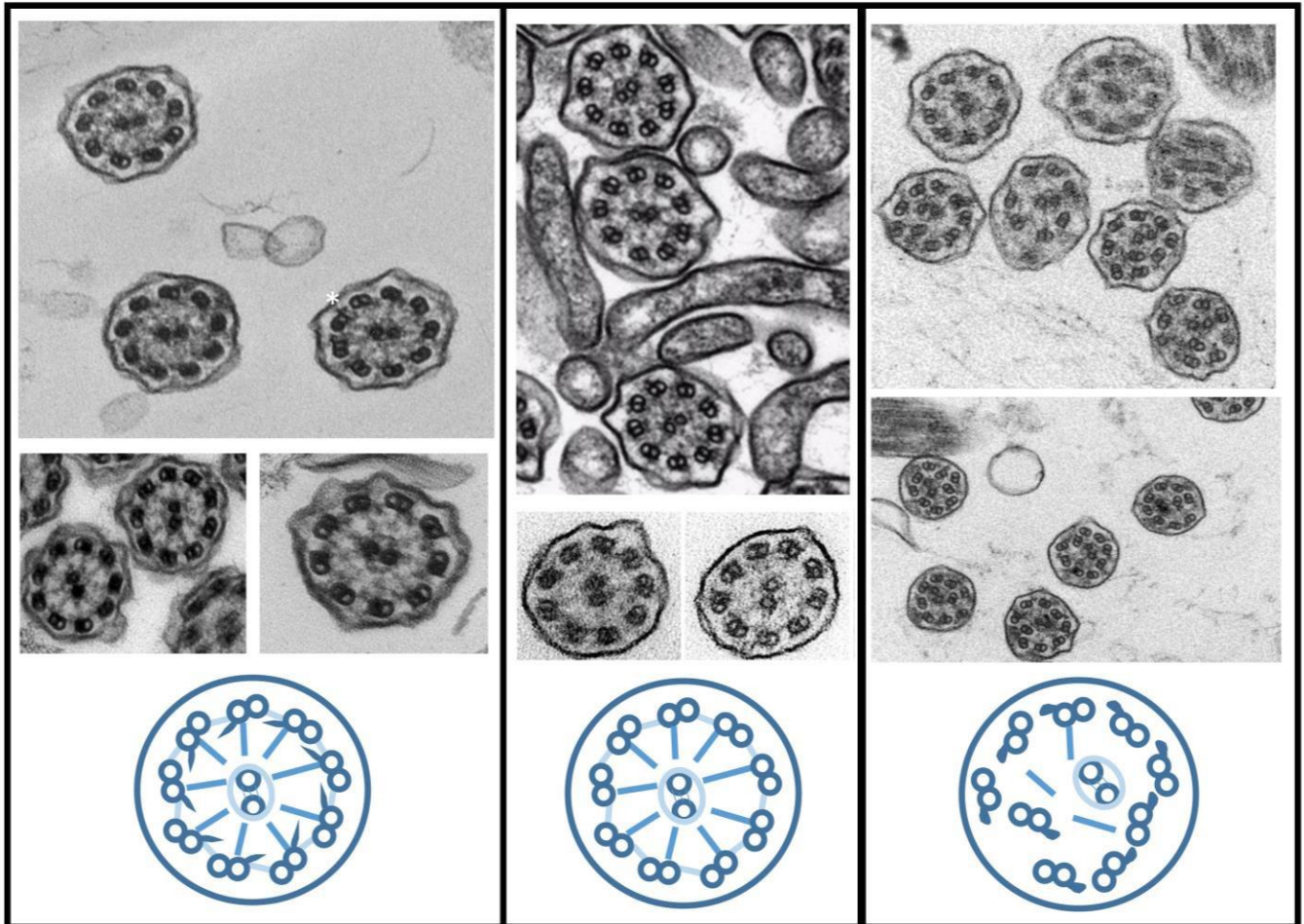
International consensus guideline for reporting transmission electron microscopy results in the diagnosis of Primary Ciliary Dyskinesia (BEAT PCD TEM Criteria)

## Normal ultrastructure of respiratory cilia.



# Class 1 defects

Defects which are considered 'hallmark' defects confirming a diagnosis in a patient with symptoms of the condition



Outer dynein arm defect

Outer and inner dynein arm defect

Microtubular disorganisation and inner dynein arm defect

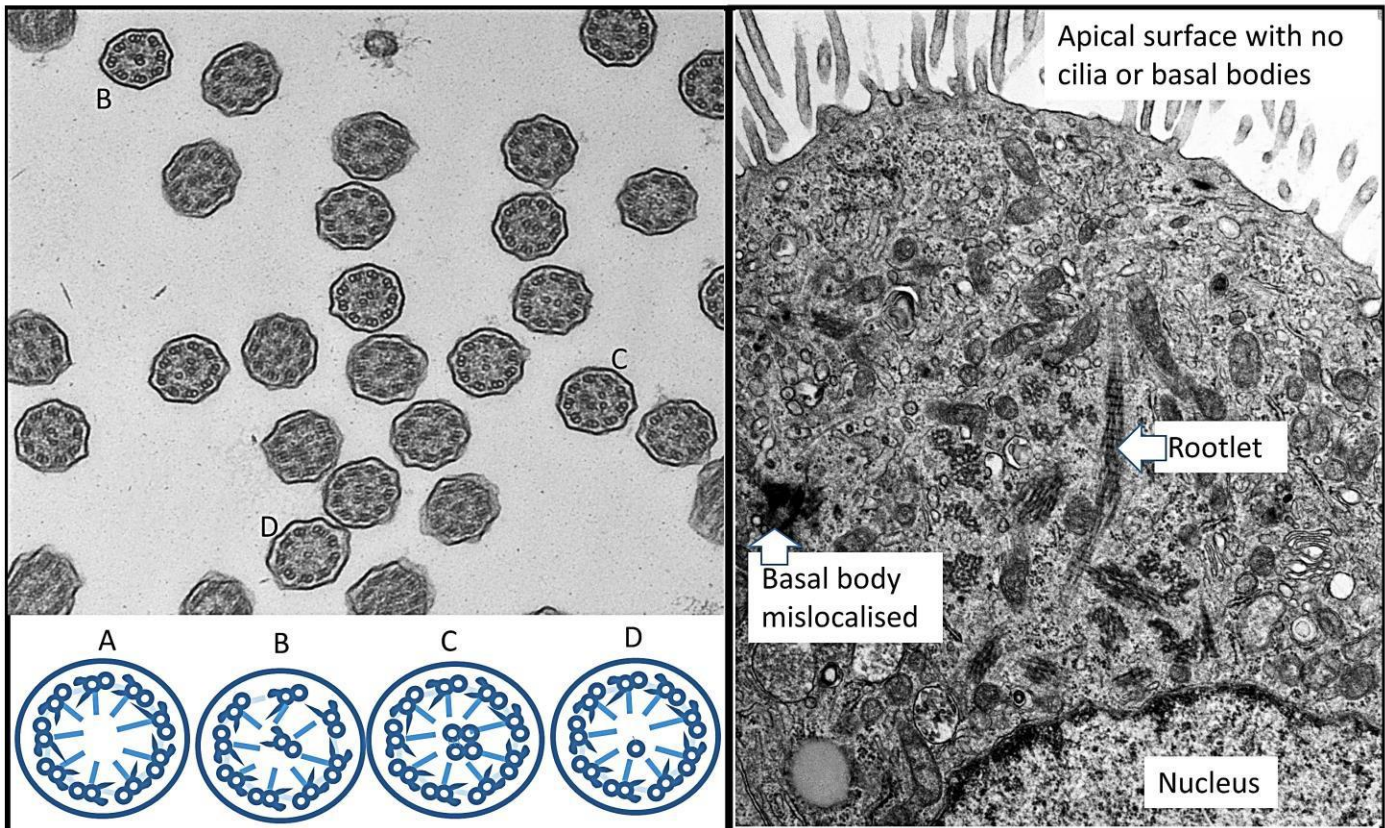
## Class 1 defects: Hallmark diagnostic defects

- Outer dynein arm defect
- Outer and inner dynein arm defect
- Microtubular disorganisation and inner dynein arm defect

*"If the number of inner dynein arms cannot be accurately determined samples should be reported as outer dynein arm defect (+/- inner dynein arm defect)"*

# Class 2 defects

Defects which indicate a diagnosis of PCD in a patient with clinical symptoms of the condition if consistent across more than one sample or after cell culture AND consistent with other test results



Central complex defect

Mislocalisation of basal bodies with few or no cilia

**Class 2 defects: Indicate a PCD diagnosis with other supporting evidence**

- Central complex defect
- Mislocalisation of basal bodies with few or no cilia
- Microtubular disorganisation defect with innerdynein arm present
- Outer dynein arm absence from 25%-50% cross sections
- Combined inner and outer dynein arm absence from 25-50% cross sections

# REPORT

## Items to include in a TEM report for the diagnosis of Primary Ciliary Dyskinesia

### Essential items to be included in a TEM report for PCD

- Source of the sample (e.g. nasal brushing)
- Adequacy of the sample
- Number of cross sections assessed
- % abnormal cilia (class 1 and class 2 defects)
- Consistency of a defect across several cells
- One sentence summary of key findings (including class 1 or 2 defect if present)

### Additional items to consider enhancing a report

- Orientation/ alignment of the basal body or central pair of microtubules
- Number of cells assessed
- Blebs/membrane swelling/membrane condition
- Presence of compound cilia (more than one axoneme within a membrane)
- Preservation of the sample
- % cilia with other defects
- Presence of shortened or truncated ODA projections
- Evidence of inflammation
- Evidence of bacteria

## **Adequacy of the sample/analysis:**

*(...)we recommend assessment of at least 50 axonemes in transverse section from a number of different cells. We recommend dynein arms should be assessed in cross sections with clear structural features and intact ciliary membranes. Microtubular arrangement **may be assessed in a larger number of cross sections with intact membranes** in which the dynein arms are not clear(...). For class 2 defects we recommend assessment of **more than one sample or following cell culture**. Paramount is the consistency of a class 2 defect with results of other tests such as high speed video, immunofluorescence analysis or genetics*