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International consensus guideline for reporting transmission electron microscopy results in the diagnosis of primary ciliary dyskinesia (BEAT PCD TEM Criteria)

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This tested and validated consensus statement provides clear terminology and defect definitions for the ultrastructural diagnosis of primary ciliary dyskinesia (PCD). The guideline will facilitate standardisation of PCD research and diagnosis. <http://bit.ly/2RDkz7k>

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ABSTRACT Primary ciliary dyskinesia (PCD) is a heterogeneous genetic condition. European and North American diagnostic guidelines recommend transmission electron microscopy (TEM) as one of a combination of tests to confirm a diagnosis. However, there is no definition of what constitutes a defect or consensus on reporting terminology. The aim of this project was to provide an internationally agreed ultrastructural classification for PCD diagnosis by TEM.

A consensus guideline was developed by PCD electron microscopy experts representing 18 centres in 14 countries. An initial meeting and discussion were followed by a Delphi consensus process. The agreed

guideline was then tested, modified and retested through exchange of samples and electron micrographs between the 18 diagnostic centres.

The final guideline a) provides agreed terminology and a definition of Class 1 defects which are diagnostic for PCD; b) identifies Class 2 defects which can indicate a diagnosis of PCD in combination with other supporting evidence; c) describes features which should be included in a ciliary ultrastructure report to assist multidisciplinary diagnosis of PCD; and d) defines adequacy of a diagnostic sample.

This tested and externally validated statement provides a clear guideline for the diagnosis of PCD by TEM which can be used to standardise diagnosis internationally.