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Title: Primary and secondary ciliary dyskinesia: Clinical phenotypes and their correlation with ciliary ultrastructural analysis

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Body: Ciliary dyskinesia (CD) is a disorder in ciliary beating that can either be primary (PCD) or secondary (SCD). The aim of this study is to identify CD phenotypes by the correlation between clinical picture and ciliary ultrastructural analysis. METHODS We have retrospectively recruited 86 patients that have undergone ultrastructural analysis for a suspicion of PCD. RESULTS A total of 108 brushing have been analysed, of which 37 with PCD, 56 with SCD, 5 with normal ultrastructure (nEM). No differences were found in age at the diagnosis. Children with nEM had a mean % of pathologic cilia of 8%, in respect to 41% in the SCD and 77% in the PCD group ($p<0,0001$). The ultrastructural abnormalities between the children with PCD, SCD and nEM showed a significant difference not only in the % of pathologic cilia, but also in dynein arm, peripheral pair and microtubuli defects. Situs inversus, respiratory distress, rhinitis, “wet” cough and bronchiectasis were significantly more seen in PCD ($p<0,05$). CONCLUSIONS Classical PCD, easy to diagnose by TEM, should be able to be identified in an early, in order to preserve lung function. The overlap of PCD and SCD in the symptoms caused by altered mucociliary clearance, and in the ultrastructural analysis makes diagnosis difficult. PCD is likely to include milder phenotypes that may be manifested by subtle or no apparent structural defects and ciliary dysfunction. Obviously the diagnosis of PCD is not only based on TEM results but includes also beating pattern and frequency analysis. If the suspicion of PCD is high, further testing should be done in order to verify the diagnosis, such as nNO, IF of ciliated cells and genetic analysis.