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Resumo	Primary Ciliary Dyskinesia (PCD) is underdiagnosed in Brazil. We enrolled patients from an adult service of Bronchiectasis over a two-year period in a cross-sectional study. The inclusion criteria were laterality disorders (LD), cough with recurrent infections and the exclusion of other causes of bronchiectasis. Patients underwent at least two of the following tests: nasal nitric oxide, ciliary movement and analysis of ciliary immunofluorescence, and genetic tests (31 PCD genes + CFTR gene). The clinical characterization included the PICADAR and bronchiectasis scores, pulmonary function, chronic Pseudomonas aeruginosa (cPA) colonization, exhaled breath condensate (EBC) and mucus rheology (MR). Forty-nine of the 500 patients were diagnosed with definite (42/49), probable (5/49), and clinical (2/49) PCD. Twenty-four patients (24/47) presented bi-allelic pathogenic variants in a total of 31 screened PCD genes. A PICADAR score>5 was found in 37/49 patients, consanguinity in 27/49, LD in 28/49, and eight PCD sibling groups. FACED diagnosed 23/49 patients with moderate or severe bronchiectasis; FEV1≤50% in 25/49 patients, eight patients had undergone lung transplantation, four had been lobectomized and cPA+ was determined in 20/49. The EBC and MR were altered in all patients. This adult PCD population was characterized by consanguinity, severe lung impairment, genetic variability, altered EBC and MR.
Fomento	