A case of patient has been diagnosed primary ciliary dyskinesia and amyloidosis

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Introduction: Primary ciliary dyskinesia(PCD) is characterized by congenital impairment of mucocilliary clearance, leading to mucus retention in the respiratory system. Amyloidosis refers to the extra-cellular tissue deposition of fibrils. We report one case who have both PCD and amyloidosis although both of them are rare diseases respectively.

Case presentation: A 33-years old female was transferred for lung transplantation from other hospital due to recurrent respiratory infection and progression of dyspnea. The patient had been diagnosed for diffuse panbronchiolitis clinically 20 years ago. She was admitted for further evaluation of dyspnea including biopsy and pre-lung transplantation evaluation. Because she had been suffered from not only respiratory symptoms such as chronic cough, sputum, dyspnea and recurrent infection but also nasal symptom including rhinorrhea and nasal obstruction since childhood. we suspected congenital diseases such as PCD and we performed nasal biopsy and lung biopsy through transbronchial lung biopsy(TBLB) . In tissue taken from nasal biopsy, there is defect with microtubular disarrangement with some central pair defect, suggestive of PCD. The tissue taken from TBLB shows deposition of non-branching fibrils in interstitium, consistent with amyloidosis and similar pattern of cilia with nasal biopsy. The echocardiography for pre-op evaluation of lung transplantation shows left ventricle wall thickness, therefore we take heart MRI and, cardiac biopsy. Heart MRI shows marked increased native T1 value, suggestive of cardiac amyloidosis. In cardiac tissue, there is amorphous material deposition in interstitium, consistent with cardiac amyloidosis. We follow up this patient and consider both of lung and heart transplantation.

Conclusion: Comorbidity of PCD and amyloidosis is very rare, however sometimes two diseases can exist simultaneously. Therefore, clinicians should exam carefully in patients who have respiratory symptoms for long time since childhood and severe rare disease can develop at the same time.

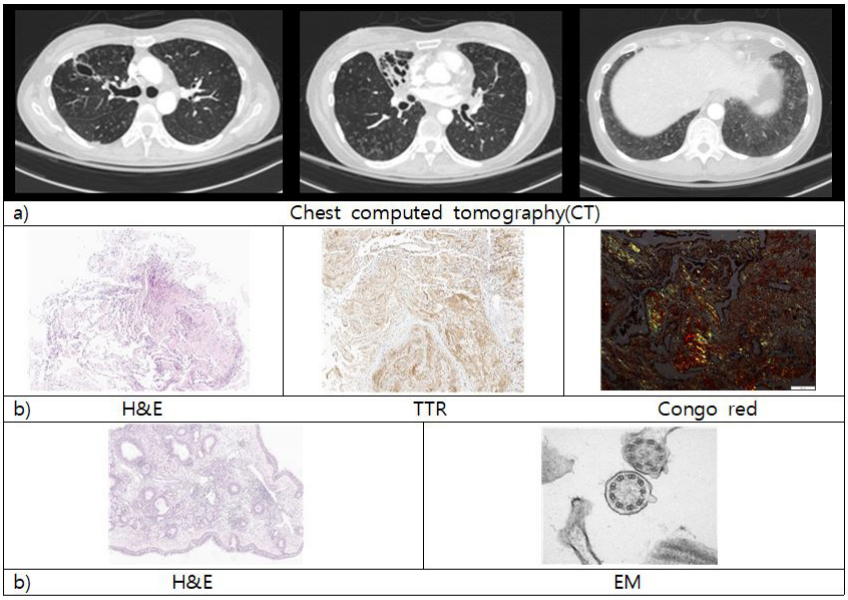

Figure 1 (a) Chest computed tomography(CT) scan showed diffuse panbronchiolitis, multifocal bronchiectatsis with mucus stagnation, GGO patterns in both lower lobes. (b) Pathologic findings from hilar lymph node, H&E – amorphous material deposition in interstitium, TTR – weak positive staining in amorphous material, Congo red – pinkish staining with apple-green birefringence. (c) Pathologic findings from nasal mucosa, H&E - Chronic nonspecific inflammation, TEM - inner dynein arm defect with microtubular disarrangement with some central pair defect, suggestive of primary ciliary dyskinesia.