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Case Series of Adult Kartagener Syndrome

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Objectives: Primary ciliary diskynesia(PCD) is a rare autosomal recessive disease and associated with ciliary functional and structural abnormalities. Approximately 50 percent of this syndrome have situs inversus totalis, chronic sinusitis and brochiectasis which is known Kartagener's syndrome. Mutations in the genes encoding the axonemal structure and accesory components of cilia can result in PCD. Infertility, recurrent rinosinusitis and pulmonary tract infections arise due to ciliary funtional abnormalities. Hyperinflation and bronchiectasis especially in middle-lower and lingular lobes are seen on high resolution computerized tomography (HRCT). Nasal nitric oxide (nNO) measurement, examination of ciliary function and structure by electron microscopy or high-speed videomicroscopy, cell culture and genetic analysis are diagnostic procedures. More than 30 different PCD causing genetic variants have been described (DNAH5,DNAH9,DNAI1, etc.). DNAI1 gene codes for the intermediate chains of outer dynein arms. Recurrent infections worsens the prognosis in this group of patients. The aim of the study is to investigate DNAI1 mutation in adult Kartagener Syndrome and to compare the clinical findings of the patients with annual exacerbation count.

Methods: Patients that have been diagnosed as Kartagener Syndrome and is still under routine follow-up in Çukurova University Department of Chest Diseases between 2013-2019 enrolled in this study. Demographic characteristics, annual exacerbation counts and the pulmonary function test results of patients at diagnosis and the end of the follow-up period recorded. And also, sputum cultures and DNAI1 mutations of the patients have been recorded

Results: The mean age of the enrolled 8 patients was 30±13.8 (22-64) and 5 of them were female. The mean follow-up period was 4.4±1.9 (0.5- 6) years. The annual exacerbation count of overall group was 1.9±1.2(0.44-4.30). The demographic and clinical characteristics of the patients. Over 50% of patients had 2 or more annual exacerbation counts. Basal FEV1/FVC and FEV1 of the patients who had rare (<2) exacerbations was higher than the frequent (≥2) exacerbated group. The FEV1/FVC and FEV1 of the frequent exacerbated group showed a greater decrease while these measurements were nearly stable in rare exacerbated group. But the difference was not statistically significant mainly due to the small sample size. DNAI1 gene mutation was negative in 2 of the patients.

Conclusion: The adult patients with Kartagener Syndrome who have a lower basal pulmonary function test results had more frequent annual exacerbations and a greater decrease in pulmonary function tests.

Keywords: Kartagener syndrome, genetics, respiratory function tests