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Current Diagnostic Methods in Primary Ciliary Dyskinesia: Hacettepe University Experience

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Objectives: Primary ciliary dyskinesia (PCD) is genetically heterogenous disease characterized by congenital abnormalities in both structure and function of the motile cilia and predominantly inherited as an autosomal recessive trait. Disease severity related with genetic analysis have been described previously in some studies including few PCD genes. The main aim of our study was to describe clinical characteristics and laboratory findings of patients with PCD, in relation to diagnostic tests including nasal NO, high speed videomicroscopy(HSVM), transmission electron microscopy (TEM) and genetic analysis. We also aimed to investigate the correlation between clinical, radiological, laboratory findings and diagnostic tests including nasal NO, HSVM and genetic analysis of these patients.

Methods: We analyzed the clinical characteristics, laboratory findings and genetic results of the 61 patients diagnosed with PCD according to history, clinical, radiological findings, nasal NO, TEM, HSVM and genetic results between January 2013-December 2018.

Results: The mean diagnostic age of the patients was 8.3±4.2 (6 months-15 years) years and initial symptoms started meanly 8 (1 month-11 years) months old. According to symptoms: 81.2% of patients have neonatal respiratory distress, 96.7% of patients have rhinitis, 82% of patients have recurrent sinusitis, 36.1% of patients have recurrent otitis, 16.4% of patients have hearing problems, 18% of patients have clubbing, 29.5% of patients have situs inversus totalis. Mean BMI was 18.8±3.6 and z score was-0.38 in the whole cohort. Mean FEV1 was 77%, FVC was 81%, FEF25-75 was 63% and z scores were FEV1, FVC, FEF25-75 -2.19, -1.87, -2.55 respectively. FEV1 (r:0.52), FVC (r:0.43) and FEF 25-75 (r:0.46) z score had significant moderate positive correlation with BMI z score. Genetic analysis revealed DNAH5 (n=11), CCDC40 (n=9), RSPH4A (n=5), DNAH11 (n=5), HYDIN (n=5), CCNO (n=4), DNAI1 (n=2), ARMC4 (n=2), TTC25 (n=1), DNAH1 (n=1), CCDC39 (n=1) and new candidate (n=15) PCD genes. According to genetic results mean diagnostic age was low in patients with CCNO mutations due to early symptoms in these patients, mean BMI and BMI z score was low in patients 17.8, 15.9 and -0.65, -0.99 respectively with CCDC40 and CCNO mutations compared with other mutations. Mean FEV1%, FVC%, FEF25-75% was 65, 73, 47 and FEV1, FVC, FEF25-75 z score -2.09, -1.81, -3.9 respectively were also lower in CCDC40 mutation group.

Conclusion: Our cohort showed that growth and nutrition are associated with lung functions in PCD patients. Patients who had mutations of CCDC40 and CCNO presented earlier and had worse lung disease and poorer nutritional status.In the future, genotype-phenotype relationships will be identified in PCD and we hope that will define the influence of specific genetic defects on lung disease.

Keywords: Primary ciliary dyskinesia, genetic, lung function test