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TERT Mutation Analysis in IPF Patients: A Pilot Cohort Study

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Objectives: Idiopathic pulmonary fibrosis (IPF) is a chronic,progressive, fibrotic disease with a poor prognosis. The exact etiology is still unknown although many factors as genetic factors, gastroesophageal reflux disease, male sex, occupational exposure, cigarette smoking have been deemed. Our aim in the study was to investigate the relationship between IPF and human telomerase reverse transcriptase (TERT) mutation.

Methods: In this study, 23 İPF patients and 22 healthy controls who admitted to Çukurova University Chest Diseases Department between October 2017 and December 2018 have been enrolled. Detailed sociodemographic and clinicinal data of all participants is recorded after acceptance of an informed consent that is approved by Cukurova University ethical committee. The blood sample of all participants have undergone some DNA isolation and analyzed for TERT mutation with PCR technique.

Results: The demographic and clinical characteristics of IPF patients and control group is shown. Only 4 (17%) of IPF patients were female and the mean age was 63.9±8.25 years. 14 (63.6%) of the control group were female and the mean age was 64.0±11.7 years. The mean age and gender was similar in both groups. The PCR analysis results of the IPF patients revealed 9 (39.1%) homozygous, 10 (43.5%) heterozygous and 4 (17.4%) normal alelles for TERT mutation. In control group, five homozygous (22.7%), 14 (63.6%) heterozygous TERT mutation and 3 (13.7%) normal alelle were detected.

Conclusion: The study is still in progress and 24 samples from IPF patients and 26 samples from control group are under PCR analyses. To the best of our knowledge, this is the first study about genetic mutations of IPF patients in Turkey.

Keywords: IPF, mutation, TERT