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Our Patients Diagnosed with Cystic Fibrosis without Sweat Test in our Region

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Objectives: For early diagnosis of Cystic Fibrosis, which is a genetic disease come to existence with birth, sweat test is of crucial importance. Patients with high immune-reactive trypsinogen (IRT) values on heel stick performed during neonatal period were used to be diagnosed with high sweat test values. However, the sweat test kit necessary for diagnosis of the disease has not been available for a year. We planned to represent our patients who were referred to our clinic for high IRT values within scope of screening program by Ministry of Health and diagnosed with cystic fibrosis without performance of a sweat test during last 1 year.

Methods: We performed clinical follow-up of the patients who admitted to our clinic with high IRT values through weight-height measurement, blood gas analysis, blood electrolytes, nasal cultures, throat cultures and fecal fat analysis. We established diagnosis of cystic fibrosis by requesting a genetic analysis from patients who were consistent with pseudobartter syndrome and had growth in nasal and throat cultures and positive fecal fat test.

Results: Of our 10 patients diagnosed during this year; 3 were female and 7 were male. After our 10 patients were diagnosed; their siblings were also investigated and 2 more children were diagnosed with cystic fibrosis. Age at the time of diagnosis ranged between 24 days and 151 days.1st IRT range was 32-300 and 2nd IRT range was 77-250. At the time of diagnosis, 7 patients (70%) had pseudobartter syndrome. In backgrounds; 3 patients (30%) had history of meconium ileus and 3 patients (30%) had history of jaundice during neonatal period. In family histories; 4 patients (40%) had consanguineous parents and 4 patients (40%) had a sibling with cystic fibrosis. During follow-ups; 10 patients (90%) had positive fecal fat tests and 90% had growth in throat cultures (1 patient had Pseudomonas aeruginosa growth at 32 days of age). Ten patients have cystic fibrosis mutations in both alleles. Three patients (30%) were found to be ΔF508'i homozygous.

Conclusion: We have diagnosed an average of 15 patients with cystic fibrosis between 2015 and 2017; we diagnosed 10 patients with cystic fibrosis during this year because of failure to perform a sweat test. Although elevated IRT helps us for screening, it has complicated the process of diagnosis of cystic fibrosis because of failure to perform a sweat test.

Keywords: Cystic fibrosis, diagnosis of cystic fibrosis, diagnosis of cystic fibrosis without sweat test