DOI: 10.5152/TurkThoracJ.2019.107

[Abstract:0720] MS-148 [Accepted: Oral Presentation] [Pediatric Lung Diseases]

Characteristics of Cystic Fibrosis Patients Diagnosed After False Negative Cystic Fibrosis Newborn Screening Results

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Objectives: The aim of Cystic Fibrosis Newborn Screening (CFNBS) is to diagnose asymptomatic infants with cystic fibrosis (CF) and to start early treatment. Since January 1, 2015, newborn screening program has been implemented in our country. However, CF can be diagnosed in patients with negative neonatal screening result. The aim of this study is to report the clinical and genotype characteristics of patients who were diagnosed as CF after false negative CFNBS results since January 2015.

Methods: Newborn screening and CF Diagnosis in Turkey
1. Blood spot sample: IRT assay (>90ng/mL)
CF suspected
2. Repeat blood spot: IRT assay (>70ng/mL)
CF suspected
Sweat test for definitive diagnosis
Normal: Cl<30mmol/L
Intermediate: 30-59 mmol/L
CF: ≥60 mmol/L

Results: 63 patients were diagnosed as CF in our clinic between 2015-2018. While 84.2% (n=53) of the patients were diagnosed by positive CFNBS result, 15.8% of the patients (n=10) had false negative results of IRT values. Of the 10 patients included in the study, 50% were girls, with a mean age of 30.7±8.8 months (12-39 months); The mean age of diagnosis was 152±147 days (28-480 days). Genetic mutation was detected in all patients. The first IRT level was above 90 µg/l in 40% (n=4) of the patients. 20% of the patients had respiratory symptoms; Respiratory symptoms and diarrhea were present in 10% and growth failure was observed in 30% of the patients. 30% of the patients were presented with meconium ileus and 10% of the patients were diagnosed due to history of CF in their brother or sister. 40% of the patients had pancreatic insufficiency and Pseudobartter was detected in 70% of patients. Pseudomonas aeroginosa growth in sputum culture was detected in 60% of patients whereas MRSA growth was 10%. 30% of the patients had anemia.

Conclusion: Regardless of the results of the newborn screening tests, patients with symptoms supporting CF should be further evaluated. For the patients with intermediate sweat test results, genetic analysis should be performed for CF. As seen in our study, it should be kept in mind that IRT negativity may also occur in mutations that cause severe CF.

Keywords: Cystic fibrosis, screening, false negativity, genetic