DOI: 10.5152/TurkThoracJ.2019.197

[Abstract:0253] OP-055 [Accepted: Oral Presentation] [Clinical Problems - Other]

Identification of Patients Diagnosed with Cystic Fibrosis Newborn Screening: Four Year Experience of A Single-Center

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Objectives: Cystic fibrosis (CF) is an autosomal recessive multisystem disorder which has negative impact on life span and quality. Newborn screening intends to diagnose infants in the first few weeks of life before the appearance of clinical signs. This study aimed to call attention to CF, a disease which can be detected with newborn screening before irreversible nutritional and respiratory system manifestations occur.

Methods: This retrospective study evaluated records of infants referred to our center due to positive CF newborn screening [immune reactive trypsinogen (IRT)] between January 2015–January 2019. Patients missing an indicated second sweat chloride test were not included. Clinical data, sweat chloride test and cystic fibrosis transmembrane regulator (CTFR) gene mutation analysis results were searched.

Results: A total of 229 infants were referred during the study period. Infants with an initial sweat chloride test reflecting<10 mmol/L (n=33) or inadequate material (n=12) were excluded if second test was unavailable. The study group consisted of 184 infants [female/male 113/71 (61.4%/38.6%)]. The median admission time was postnatal 26 (9-98) days. The mean gestational week was 386±1.75 and mean birth weight was 3300±432 gr. All infants passed their first stool in 48 hours at most. Five infants (3.6%) faced prolonged jaundice two (1.4%) had neonatal pneumonia and 2 (1.4%) had coughing. Parental consanguinity was positive in 12 (8.6%) infants. Weight gain was insufficient in seven (5%) infants. Sweat chloride test was normal in 154 (83.7%) infants, ≥60 mmol/L in one infant and ≥30-<60 mmol/L in eight (4.3%) infants. Second test was administered in thirty-one infants. Two out of fifteen infants with an inadequate material in the initial test reflected high second test results. All six infants with an initial test<10 mmol/L had a normal second test. Repeated sweat chloride test was high in six infants. One of these patients had prolonged jaundice while one had neonatal respiratory distress. CTFR mutation analysis of these infants revealed one homozygote F508 deletion and three 5T/7T/9T polymorphisms. Treatment was initialized in four patients while two were conservatively followed-up.

Conclusion: Cystic fibrosis is an inherited disease with a higher prevalence in countries like ours possessing frequent consanguineous marriage. Newborn screening is effective against the prevention of progressive lung injury and malnutrition. In addition, advanced treatment modalities have extended life span in CF. The dissemination of CF newborn screening, also held in our country, will provide better outcome concerning the morbidity and mortality of these patients in the future.

Keywords: Cystic fibrosis, newborn screening, sweat chloride test