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A Mutation Leading to Atypical Presentation of Cystic Fibrosis: D1152H CFTR Mutation and Clinical Characteristic of the Patients

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Objectives: Cystic fibrosis is the most common multisystemic disease exhibiting an autosomal recessive inheritance pattern in white race. In children with cystic fibrosis that carry some mutations including D1152H, may have sweat chloride levels which are at borderline or within physiological range. Our objective is to provide some information about the presentation of this genetic mutation.

Methods: Clinical presentations of 10 patients with detected D1152H mutation who were being followed-up with diagnosis of cystic fibrosis in Department of Pediatric Pulmonology of Necmettin Erbakan University Meram School of Medicine were represented.

Results: Of the patients with detected D1152H mutation; 6 were female and 4 were male. 6 patients were diagnosed during the process of genetic counseling due to a family history of cystic fibrosis and 4 were diagnosed with a complaint of chronic sinopulmonary infection. 9 of the patients were familial cases. 3 patients were cousins of 4 patients (at 10, 12, 16 and 21 years of age), the other 2 patients (at 4 and 9 years of age) were siblings. Patients' median age was 5.4 years (1 month – 25 years) and median sweat test value was 62.4 (32.5-103) mmol/L. Only 1 patient had a value over 90 mmol/L, which is the cut-off value for diagnosis. Bronchiectasia was present in 40% of the patients. Colonization with Pseudomonas aeruginosa was present only in 3 of 10 patients. 8 (%80) of 10 patients were found to have a FEV>80%. Annual rate of reduction in FEV 1 was lower compared to age-matched patients with classical CF mutations. However, when D1152H mutation coexisted with another mutation leading to cystic fibrosis, pulmonary symptoms were determined to be more prominent. Only 1 patient required hospitalization during last 1 year. Idiopathic pancreatitis developed in 5 patients. Fecal fat test was positive in all patients. 2 patients were found to have a gallstone. 6 patients had elevated liver enzymes. BMIs of 4 patients older than 16 years of age were >15. Z scores of other 6 patients were within physiological range.

Conclusion: Results of our study suggest that patients' clinical presentation is determined by the accompanying mutation, sweat chloride levels of these patients may be at borderline or within physiological range, they have a pulmonary infection less frequently, they are diagnosed later due to subtle signs, they have less amount of P. Aeruginosa colonization, it should be considered in patients with idiopathic pancreatitis and there is a long survival.

Keywords: Atypical mutation, child, D1152H, cystic fibrosis