Clin Pediatr OA 2018, Volume 3
DOI: 10.4172/2572-0775-C2-012

Normal sweat chloride test does not rule out cystic fibrosis

Abdurrahman Erdem Başaran
Akdeniz University, Turkey

Cystic fibrosis is an autosomal recessive disease, which results from a mutation in the transmembrane conductor regulator gene. Nowadays, measurement of chloride level in sweat (sweat test) is the most widely used biochemical method for the diagnosis of cystic fibrosis. However, there are various reports in literature, regarding false positive or false negative sweat test results in CF patients. Therefore, the interpretation of sweat test results should be done by taking into consideration the clinical condition of the patient and possible accompanying comorbid situations. One of the reasons leading to the negative results on sweat test is the presence of specific mutations in CF patients. In literature, we report a five-month-old boy who was diagnosed as CF with the findings of pseudo-Bartter's syndrome and compound heterozygous for p.E92K/p.F1052V mutations, but with negative for sweat test. Other causes of the false negative results on sweat test are indicated as technical reasons, specific mutations, hypohidrotic ectodermal dysplasia, inadequate sweat collection, mineralocorticoid treatment, young age, edema, hypoproteinemia and penicillin treatment. The first four reasons are the most common reasons and this specific mutation maybe responsible for our negative sweat test result. In conclusion although the sweat chloride test is the most commonly used method for the diagnosis of cystic fibrosis, it does not always give a clear answer. Mutation analysis may be helpful when clinical findings are suggestive of cystic fibrosis, even if sweat test results are negative.

erdembasaran15@hotmail.com