عنوان مقاله:

Analysis of CFTR Gene Variants and clinical presentations in Children with Diffuse Bronchiectasis and Unknown Etiology

محل انتشار:

مجله بین المللی کودکان, دوره 9, شماره 2 (سال: 1400)

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خلاصه مقاله:

Background: Diffuse bronchiectasis is an irreversible abnormal dilation of proximal subsegmental bronchi. The aim was to investigate and compare CFTR gene mutations and clinical presentations in children with idiopathic bronchiectasis. Materials and Methods: In a cross-sectional study, all children with idiopathic bronchiectasis who were hospitalized from ۲۰۱۹ to ۲۰۲۰ in Tabriz Children's Hospital, Iran, were reviewed. Bronchiectasis confirmation was based on signs, symptoms, and HRCT findings. Data was collected through medical records, medical history, clinical examination, and para-clinical examination. CFTR variants were examined by liquid chromatography, direct sequencing, and multiple probe ligations. Then children were divided into two groups based on variants identified in the CFTR gene and compared in terms of demographic, clinical, and para-clinical findings. Descriptive statistics, Chi-square Tests, and independent samples t-test was used to analyze the data using SPSS software version ۲۲.۰. Results: Out of ۲۱ patients, ۰ (۴۷.۶%) children were males with a mean age of ۹.۷۵ years. Out of ۲۱ children with diffuse bronchiectasis, five clinically significant CFTR-related gene variants were identified (group ۱). Other patients either had only single polymorphism or no variants related with CFTR (group ۲). Age, FEV۱ and sweat test were lower in group ۱ than in group ۲. Conclusion We observed the CFTR variants in heterozygote form in children with diffuse bronchiectasis with a normal or borderline sweat test. Therefore, it is necessary to determine whether DB is a part of CFTR-Related Diseases failing to meet the diagnostic criteria of Cystic fibrosis or a disease independent of Cystic fibrosis.

کلمات کلیدی:

Children, CFTR varients, Cystic fibrosis Diffuse bronchiectasis