Clinical and Genetic Characteristics of Children with Cystic Fibrosis in Henan China

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Abstract

Background: Despite the growing awareness of Cystic Fibrosis (CF) in China, few cases have been reported in Henan, which is the most populous province in the country. This study aimed to describe the clinical phenotype and genotype of children with CF in Henan. Methods: We recruited 14 Chinese children with CF who presented to Children's Hospital affiliated to Zhengzhou University from January 2019 to January 2023. The demographic data, imaging examinations, and laboratory tests of the patients were reviewed to clarify the clinical phenotype. Whole exome sequencing was conducted to identify the genotype. Results: Respiratory diseases were the main clinical manifestation, including recurrent/persistent pneumonia (85.7%), sinusitis (71.4%), bronchiectasis (71.4%). CF-related liver disease (CFLD) and pancreatic insufficiency (PI) were less common (21.4% each). Infant cases had high frequency of pseudo-Bartter Syndrome (80.0%). Chest computed tomography showed bronchiectasis in older children and air trapping in infant cases. The most common pathogens in the airway were Pseudomonas aeruginosa and Staphylococcus aureus (71.4% respectively). Twenty-four different CFTR gene variants were detected, including four novel observations (c.869+3A>T, c.1064C>G[p.Pro355Arg], c.1209+1G>C and c.1925C>G [p.Ser642X]). The most common variant was c.2909G>A (p.Gly970Asp), with a detected rate of 16.7%. Conclusion: Children with CF in Henan had varied clinical phenotypes by age, with respiratory disease being predominant. The most frequent CFTR gene variant was c.2909G>A(p.Gly970Asp). This study is the first and most comprehensive one on the clinical phenotype and genotype of children with CF in Henan, China. We also reported the first CF case of M. abscessus infection in China.

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