Gene Mutations for Cystic Fibrosis and Predisposition to Chronic Rhinosinusitis in Chinese Children

Yang Han[1], Pengpeng Wang[1], Wenjing Liu[1], Xiao Xiao[1], Xiaojian Yang[1], Lixing Tang[1], Wei Zhang[1], JIE ZHANG[1], Wentong Ge[1], and Xin Ni[1]

[1]Beijing Children's Hospital Capital Medical University

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Abstract

Background: Chronic Rhinosinusitis (CRS) is a common disease in children. In recent years, a new airway epithelial ionocyte has been discovered and is closely related to the expression of cystic fibrosis transmembrane conductance regulator (CFTR), which provides new ideas for the study of the mechanism of CRS. However, the carrying status of the CFTR mutant gene in the Chinese population is not clear. Objective: To study the frequency and mutations of CFTR and FOXI1 (ForkheadBoxI1) in Chinese children with CRS, and to analyze whether they are predisposed to CRS. Methods: A controlled case study was conducted from 2020 to 2021. The CFTR and FOXI1 genomes of 46 children with CRS and 23 children with no history of CRS from the Chinese mainland area were sequenced, and the relationship between mutation rate and the disease were analyzed. Results: 13 CFTR gene mutation sites' carrying rate in the CRS group was higher than the control group, and 2 of them was significantly higher than that in the East Asian population database. The children who had a history of recurrent upper respiratory tract infection carried CFTR gene mutations associated with the pathogenesis. There was no difference in the carrying of FOXI1 mutation between the two groups. Conclusion: The incidence of CF mutation is higher in Chinese children with CRS who have no history of Cystic Fibrosis (CF). c.650A> G (p.E217G) and c.1950C> A (P. F 650 L) and may play a role in the development of CRS conditions in children in China.

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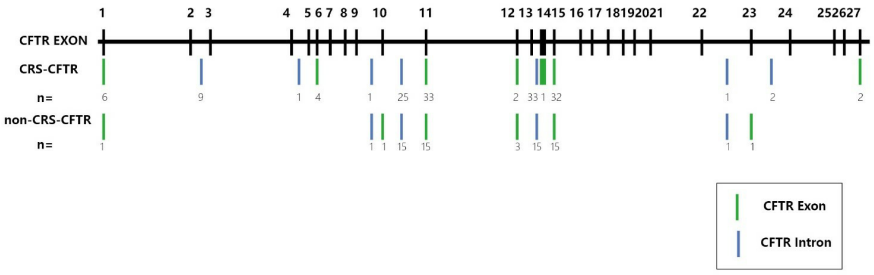

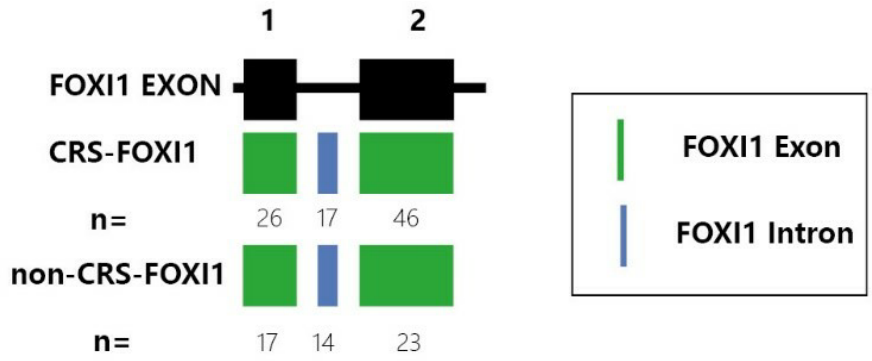

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