Cystic fibrosis transmembrane conductance regulator gene screening and clinical correlation in Taiwanese males with congenital bilateral absence of the vas deferens

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摘要

Abstract

BACKGROUND: In Taiwan, an area with a very low incidence of cystic fibrosis (CF), we first screened for the most common mutations of the cystic fibrosis transmembrane conductance regulator (CFTR) gene and looked for clinical correlations in 27 patients with clinically diagnosed congenital bilateral absence of the vas deferens (CBAVD). METHODS AND RESULTS: The clinical results showed that none of the 27 patients had CF symptoms. We did not detect any definite renal anomaly ultrasonographically. Mutation analysis was carried out on these 27 cases and 46 normal fertile males as controls. No mutations of F508 or R117H were identified in any of the samples analysed. In the screening of IVS8-poly T, five of the 27 CBAVD patients showed the homozygous genotype for 5T/5T, 14 showed the heterozygous genotype for 5T/7T and eight showed the homozygous genotype for 7T/7T. The frequency of 5T alleles was 44.4%, which was significantly higher than in the 46 normal fertile males, for which there was a 5T frequency of 5.4%. CONCLUSIONS: The absence of major mutations of CFTR genes could be related to the much lower CF incidence in Taiwan. Further investigations into differences in the mutation spectrum of other CFTR genes are needed for a better understanding of the development of Taiwanese-Oriental CBAVD.