Abstract

Introduction: Cystic fibrosis (CF) is the most frequent autosomal recessive disorder in Caucasian population with an incidence of 1 in 2000 newborns. The disease is caused by mutations in the cftr gene, but the most common mutation is F508del, which accounts for 66% of CF chromosomes worldwide and a carrier frequency for Caucasian population of 1 in 25. Objective: To determine the carrier frequency of the F508del mutation in 110 unrelated, healthy students from the Facultad de Medicina de la Universidad del Rosario. Methods: The presence of F508del mutation using PCR and heteroduplex analysis was determined. Results: Only four heterozygotes for F508del mutation were discovered. This represents a carrier frequency of 1 in 27 students. Conclusions: This estimated frequency of F508del carriers is higher than expected, encouraging further screening in normal control individuals from different regions of Colombia.

Keywords

Cystic fibrosis; deltaF508; Carrier; Colombia.