Association Study of IVS8TGmTn Polymorphism and Cystic Fibrosis Disease in a Tunisian Population

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Abstract

Background: Cystic fibrosis (CF) is the most common autosomal recessive disease in Caucasians, caused by mutation in cystic fibrosis transmembrane conductance regulator (CFTR). The analysis of some extra and intragenic markers within or closely linked to CFTR gene is useful as a molecular method in clinical linkage analysis. Indeed, knowing that the molecular basis of CF is highly heterogeneous in our population is explained in the present study. The goal of the study was to examine the IVS8 TGmTn genotypes in a Tunisian sample of patients with CF disease and normal controls, and to compare the results with the findings from the literature.

Methods: We conducted a case–control study in a sample composed of 80 CF patients and 90 control subjects to investigate the possible effect of the polymorphism. The study of the IVS8 TGmTn polymorphism was performed using sequencing analysis.

Results: Our data revealed an association between the IVS8 TGmTn polymorphism and CF risk. There was a significant difference in the poly-T; TG repeats allele or genotype frequencies between cases and controls. Also, statistical analysis shows no association between F508del mutation and T5 variant. Furthermore, new TG9 repeat was reported in CF patients**.**

Conclusion: This study provides additional evidence that TGmTn poly tract has important role in the development of CF. Further investigation, which genetic and functional studies of polymorphisms in genetic diseases will become of major interest in the future.

Keywords: Cystic fibrosis; IVS8 TGmTn polymorphism; *CFTR* gene; F508del mutation; Tunisian population