Original Paper

Common CFTR gene mutations in cystic fibrosis patients in Mazandaran province - Iran

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Abstract

Background and Objective: Cystic fibrosis (CF) is the most common inherited disorder in Caucasian populations caused by mutation in cystic fibrosis transmembrane conductance regulator (CFTR). The type of mutations and their distributions varies widely between different countries and/or ethnic groups. The aim of this study was to characterize mutations involved in this disease in Mazandaran province, Iran.

Materials and Methods: In this descriptive study thirty unrelated Iranian cystic fibrosis patients were screened for deltaF508, N1303K, G542X, R347H and W1282X mutations in the CFTR gene using Reverse Dot Blot method during 2004-06. This technique uses biotinilated PCR products for simultaneous hybridization with several normal and mutant probes specific to known mutations fixed on Biodyne C membranes.

Results: DeltaF508 mutation was found in 13 (21.66%) alleles. 6 patients were homozygote and one was compound heterozygote for this mutation.

Conclusion: These findings reveal an important heterogeneity of CFTR gene mutations in Mazandaran Province. Thus regarding the relative low rate of detectable mutations, it is necessary to undertake larger studies for molecular diagnosis of cystic fibrosis in this province.

Keywords: Cystic Fibrosis, CFTR, Mutation, Mazandaran, Delta F508

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