J Cyst Fibros. 2011 Jan; 10(1): 66-70.

Cystic fibrosis in Chilean patients: Analysis of 36 common CFTR gene mutations.

Lay-Son G, Puga A, Astudillo P, Repetto GM; Collaborative Group of the Chilean National Cystic Fibrosis Program.

Abstract

BACKGROUND: CFTR gene mutations have worldwide differences in prevalence and data on Chilean patients is scarce.

METHODS: We studied 36 of the most common CFTR mutations in Chilean patients from the CF National Program [Programa Nacional de Fibrosis Quística (PNFQ)] of the Ministry of Health of Chile.

RESULTS: Two hundred and eighty-nine patients were studied. Fourteen different mutations were identified with an overall allele detection rate of 42.0%. Mutations with frequencies greater than 1% were p.F508del (30.3% of alleles), p.R334W (3.3%), p.G542X (2.4%), c.3849+10Kb C>T (1.7%), and p.R553X (1.2%). A north to south geographical gradient was observed in the overall rate of detection.

CONCLUSIONS: Southern European CFTR mutations predominate in the Chilean population, but a high percentage of alleles remain unknown. Geographical heterogeneity could be explained in part by admixture. Complementary analyses are necessary to allow for effective genetic counselling and improve cost-effectiveness of screening and diagnostic tests.