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PRELIMINARY REPORT

The +1858 C/T Polymorphism in the PTPN22 Gene Is Associated with Cystic Fibrosis Patients in Northeast Mexico

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Cystic fibrosis (CF) is the most common autosomal recessive disease in the Caucasian population, but it has also been widely diagnosed in the Mexican population. Production of viscous secretions affects the secretory epithelia and the respiratory condition usually leads to death. The relationship between the CFTR genotype and the disease phenotype is not well understood. Other risk factors such as genetic and autoimmune influence the development of this disease. We analyzed the PTPN22 R620W polymorphism (+1858 C/T, rs2476601) in 78 DNA samples from CF patients and 232 healthy controls from northeast Mexico using the polymerase chain reaction-restriction fragment length (PCR-RFLP) method. The C allele and the CC genotype were the most frequently detected in controls (CC genotype 96.12%; C allele 98.06%) compared with CF patients (CC genotype 88.46%, C allele 93.59%). A statistically significant association for the CT + TT genotypes ($p = 0.012$, OR = 3.232) as well as for the mutant T allele ($p = 0.005$, OR = 3.463) was found when comparing CF patients with controls. A significant association was found between the rs2476601 polymorphism of the PTPN22 gene and CF in Mexican patients. Further studies are necessary to understand the influence of this variant on lung neutrophil function and disease development.

Key Words: Cystic fibrosis, +1858 C/T PTPN22, Autoimmune diseases, PCR-RFLP, Mexican population.

Introduction

Cystic fibrosis (CF) is the most common autosomal recessive disease in the Caucasian population with an incidence of 1/3,000 live births, although its frequency may vary in specific subgroups (1). According to the Cystic Fibrosis Database (http://www.genet.sickkids.on.ca/StatisticsPage.html), there are more than 2000 different mutations in the CFTR gene, the most common being the deletion of phenylalanine in the 508 position (60–70% of allele frequency). CF is characterized by an imbalance in the transport of water and electrolytes in the secretory epithelia of distal airways, intestine, and bile and pancreatic ducts, with the production of viscous mucus secretion (2). Deterioration is progressive, and the respiratory condition produced by chronic infection and inflammation usually leads to death between the first and fourth decades of life. In the diseased lung, neutrophils are recruited at the alveolar epithelium and release toxic oxygen metabolites and enzymes and stimulate the epithelium to produce chemotactic compounds, perpetuating the inflammatory response (3).

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