

As the deficiency of alpha-1 antitrypsin glycoprotein affects the severity of lung disease among patients with Cystic Fibrosis

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Abstract

Cystic Fibrosis (CF) is a serious genetic disease, CF is an autosomal recessive genetic disease clinical manifestations can vary being detectable after birth or in adulthood. Basically the defect affects the cells of various organs, which can be presented in different ways in individuals with the disease.

With this research can therefore conclude that the alpha-1 antitrypsin deficiency is an aggravating factor for lung disease in people with cystic fibrosis.

Key words:

alpha-1 antitrypsin, Cystic fibrosis, polymorphism

Introduction

Cystic Fibrosis (CF) is a serious genetic disorder, yet no cure, its incidence varies according to ethnic groups. Clinical manifestations can vary being detectable after birth or in adulthood. Basically the defect affects the cells of various organs, which can be presented in different ways in individuals with the disease. CF is an autosomal recessive genetic disease, which can be classified into 6 types according to their mutations, ie, how they affect the CFTR protein, and the severity of the disease. Alpha-1-antitrypsin (A1AT) is a glycoprotein mainly produced by the hepatocytes release daily approximately 2g of the protein in circulation. The main function of A1AT is to inhibit neutrophil elastase, a serine protease which has the ability to hydrolyze elastin fibers in the lungs. However, due to changes in protein structure caused by mutations in your gene, sometimes the protein loses its inhibitory ability or is added in the form of inclusion bodies in hepatocytes, leading to reduction of their normal levels. This research aims to analyze the polymorphism Alpha 1-Antitrypsin, and relate to deficiency of glycoprotein severity of lung disease in cystic fibrosis.

From the patients with cystic fibrosis blood sample, extraction of DNA Peripheral blood was taken with lithium chloride and proteinase K. After extracting the PCR procedure was done.

DNA	1
TpKCl	5
MgCl ₂	4
DNTP	4
PS	1
PAS	1
Taq	0,6
H ₂ O	34,4

Table 1: Concentration (uL) of the reagents used in PCR.

In sequence was made electrophoresis, where it was observed that the alpha -1 antitrypsin polymorphism particularly affects the severity of lung disease, it is involved in infection control. This glycoprotein may be effective in the treatment of CF, it protects the tissue from proteolytic attack by leukocyte proteases such as elastase, cathepsin and trypsin, during inflammatory reactions.

Conclusions

Therefore we conclude that the alpha-1 antitrypsin deficiency is an aggravating factor for lung disease individuals with cystic fibrosis.

Results and Discussion