α-1-ANTRITRIPSINA

PATHOGENESIS

In patients with chronic bronchitis is thought that the progressive destruction in alveolar structures occurs thanks to the imbalance, in the distal respiratory section, among proteases, enzymes, which can destroy alveolar structures and anti-protease. The alfa-1-antitripsina is an enzyme, which belongs to the family of “serine protease inhibitors”, whose function is the one to protect tissues from enzymatic digestion. Among factors, which could disturb this equilibrium (protease/anti-protease) there are on the one hand Broncho pulmonary infection and on the other hand hereditary lack and/or the smoke.

The pathogenic theory of protease and anti-protease in emphysema received a lot of support thanks to the demonstration that in patients with serious local deficits of alfa-1-antritripsina there is even a strong reduction of serum levels of alfa-1-antitritripsina and that these patients develop a panacytose, progressive emphysema. Recently, it was demonstrated that two polymorphisms on the gene of the alfa-1-antitripsina (allele S and allele Z) are involved in this pathologic process (normal population has the M form).

In a study on 266 patients it was demonstrated how the allele Z is a risky genetic factor for the COPD (chronic obstructive pulmonary disease). Moreover, people with a genetic configuration MS and MZ show a pulmonary emphysema just if beyond genetic components there is even the influence of noxious substances such as smoke or infections.

EPIDEMIOLOGY

The variant S is in about the 28% of south-Europe population and it guides to serical levels of alfa-1-antitripsina corresponding to 60% of the normal ones. The variant Z results on the other hand in an enzymatic rate corresponding the 10% just of the normal one in serum and it predisposes carrier person of the homozygote form (ZZ) to the pulmonary emphysema in youth (type panlobulare which distinguishes itself from the centribulare one of smokers), to the juvenile hepatitis, to the cirrhosis of the liver and to the carcinoma hepatic cellular. The hepatic involvement often occurs with the MZ form. About the 10% of people with ZZ form develop a hepatic disease normally present in childhood.

TEST

Research of polymorphisms S and Z by means of PCR and restriction analyses.

SAMPLE TAKING

Blood/EDTA, 5 ml.

EXECUTION

Daily.

COST

According to federal rate table of the analyses (2215.03X2) TP 310.

Laboratorio di diagnostica molecolare

Further information or bibliographic references can be asked to the laboratory.