

Associated study of polymorphic variants in the gene for superoxide dismutase (Mn SOD) and different types of diabetes mellitus

S.Pashkunova- Military Medical Academy –Sofia, Bulgaria

**Superoxid dismutase (SOD)** is a key antioxidant enzyme engaged in detoxication of superoxide radicals. Ala/Val substitution in Mn SOD leads to a change in the effectiveness of the action of the enzyme. This polymorphism we analyzed among several groups patients with diabetes and we compared with control group of healthy volunteers. It is proved an association connection of tracked marker in some of the groups of patients with diabetes compared to the rest population sample. Such a study is made for the first time in Bulgaria, and indicates that, polymorphic allele is meeting in- often in some of the groups of diabeticians, as the difference in frequencies relative to the control sample is statistically reliable

#### **Methodology :**

In this study a case-control we had examined patients with different types of diabetes mellitus and a control group of healthy persons.

The polymorphism Ala(-9)Val in Mn-SOD gene represents a substitution at position -9 leading to schematic amino acid substitution of alanine with valine. The genotyping of selected groups of patients and the control sample included amplification of area of Mn-SOD gene with primers containing nucleotide discrepancy creating place recognized of restrictase BshTI. So the amplified product with length 91 nucleitides, visualized after electrophoresis separation as two fragments of length 17 and 74 nucleotide pairs in the presence of polymorphic allele.

#### **Results :**

In comparing the allele frequencies and genotyping frequencies in the group of patients with diabetes mellitus type 1 and type 2 and the control sample of Mn-SOD polymorphisms was not established statistically significant difference.

#### **Patients with diabetes mellitus type 2**

The allele containing restriction place Val (+) is meeting emphasized in- often in the group with DM type 2, with an increase of the sampled group may be expected to reach statistical reliable difference.

Statistical analysis shows that the genotype containing allele (+ Val) in homo- or heterozygote condition compared with allele (- Ala) shows statistical significant difference in this group. It can be interpreted that polymorphic genotype is associated with susceptibility to this type diabetes.

#### **Patients with DM type 1**

Established statistically significant difference when comparing the allele frequencies in the group of patients with diabetes mellitus type 1, and a control sample of Mn-SOD polymorphisms. And in this group polymorphic allele occurs more often in the group of patients, the difference in frequencies relative to the control sample is statistically reliable.

In comparing the genotype frequencies shows that, the polymorphic genotype excel in - highly in homozygote condition and is meeting statistical in - often in patients with diabetes type 1.

### **Discussion**

No effective action of Mn SOD is following of leaving the mitochondria without to exercise full protection against superoxid radicals. This leads to increase the protein oxygenation as mitochondrial DNA mutations and damage, a frequent event in the pathogenesis of diabetic polyneuropathy.

The functional role and the possible association with diabetic pathology determined SOD genes as an interesting subject for associated studies.