Mitochondrial mutation in iranian patients with multiple sclerosis, correlation between haplogroups H, A and clinical manifestations

ABSTRACT

As multiple sclerosis (MS) has long been known to be associated with Leber, hereditary optic neuropathy (LHON), a disease caused by mitochondrial (mtDNA) mutations, in this study we assessed possible involvement of mtDNA point mutation in MS patients. Fifty-two MS patients whose disease was confirmed with revised McDonald criteria and referred to Iranian Center of Neurological Research of Imam Khomeini hospital during 2006ï 2007 entered the study. Secondary mtDNA mutations, age, gender, clinical disability according to expanded disability status scale (EDSS), course of the disease, and presenting symptoms were the variables investigated in this study. DNA purification was performed by Diatom DNA Extraction Kit. Analysis of data was done by SPSS V11.5. The prevalent mutations with frequency of 19.2% were J, L, and T haplogroups. Haplotype A was more prevalent in patients with younger age of onset (P-value = 0.012) and high proportion of haplogroup H was associated with optic nerve involvement (P-value = 0.015). No motor symptoms were seen in haplogroup H patients. There is no significant relationship between duration of the disease and EDSS in different mutation of mtDNA.

Keyword: Multiple sclerosis; Mitochondrial DNA(mtDNA) mutation