Molecular analysis of the SMN and NAIP genes in Iranian spinal muscular atrophy patients

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Spinal muscular atrophy (SMA) is an autosomal recessive neuromuscular disorder characterized by degeneration of spinal cord anterior horn cells, leading to muscular atrophy. SMA is clinically classified into three subgroups based on the age of onset and severity. The majority of patients with SMA have homozygous deletions of exons 7 and 8 of the survival motor neuron (SMN) gene. The purpose of the present study was to determine the frequency of SMN and neuronal apoptosis inhibitory protein (NAIP) gene deletions in Iranian SMA patients. Experience in prenatal diagnosis of SMA in this population is also reported. To study the frequency of deletions of SMN and NAIP genes in an Iranian sample group, 75 unrelated SMA patients (54 type I, eight type II and 13 type III) were analyzed according to the methods described by van der Steege et al and Roy et al. Homozygous deletion of SMN1 exons 7 and/or 8 were identified in 68 out of 75 patients (90%). Deletion of exon 5 of the NAIP gene was found in 40/54 of type I, 2/8 of type II and 1/13 of type III patients. Deletion of the SMN1 gene is a major cause of SMA in Iran, and NAIP gene deletions were common in the present patients with type I SMA. Also, the incidence of NAIP deletion is higher in more severe SMA.

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