Journal of Pediatric Neurology 10 (2012) 269–273
DOI 10.3233/JPN-120581
IOS Press

Correlation between deletion patterns of SMN1 and NAIP genes and clinical subtypes of spinal muscular atrophy in Iranian Azeri Turk ethnic patients

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Received 9 March 2012
Revised 30 April 2012
Accepted 17 May 2012

Abstract. The childhood spinal muscular atrophy (SMA) is classified into three groups based on the age of onset and clinical course. The aim of this study was to define the correlation between genotype and phenotype in Iranian patients with SMA. Molecular analysis was carried out on two candidate genes of survival motor neuron one (SMN1), and neuronal apoptosis inhibitory protein (NAIP) in 189 patients with SMA (130 patients with SMA type I, 32 patients with type II and 27 patients with type III) and the phenotypic features of SMA were compared with the deletion pattern of these genes. Polymerase chain reaction along with restriction fragment length polymorphism analysis were used to detect the deletion of exons 7 and 8 of SMN1 gene, as well as multiplex polymerase chain reaction for exon 5 of NAIP gene. Deletion in exon 7 was detected in 167 patients (88.4%); deletion in exon 8 was detected in 162 patients (85.7%) and deletion in exon 5 of NIAP gene was detected in 95 patients (50.3%). Deletion of exons 7 and/or 8 of SMN1 were detected in 90%, 84.4%, and 88.9% in patients with types I, II and III SMA, respectively. The prevalence rates of exon 5 deletion in NAIP gene was 66.9%, 12.5%, and 14.8% in types I, II and III SMA patients, respectively ($P < 0.001$). Combined homozygous deletion in both SMN1 and NAIP genes was found in 65.4% SMA type I, 12.5% SMA type II and 14.8% SMA type III ($P < 0.001$).There was a significant correlation between combined deletion of both SMN1 and NAIP genes and clinical subtypes of SMA patients.

Keywords: Spinal muscular atrophy, survival motor neuron gene, neuronal apoptosis inhibitory protein gene, genotype and phenotype correlation

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