The Clinical and Genetic Spectrum of six patients with Spinal Muscular Atrophy from Northern Iran

M Omran, A Juibary

Citation

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Abstract

Autosomal recessive spinal muscular atrophy (SMA) is, after cystic fibrosis, the second most common fatal monogenic disorder. The disease is characterized by degeneration of anterior horn cells leading to progressive paralysis with muscular atrophy. Depending on the clinical type (Werdnig-Hoffmann = type I, intermediate form = type II, Kugelberg-Welander = type III), SMA causes early death or increasing disability in childhood. To describe the clinical findings of patients with spinal muscular atrophy (SMA) with survival motor neuron (SMN) gene deletion.

Descriptive study of SMA cases confirmed with the deletion of the SMN gene. Frequency determination of positive clinical and laboratory revised diagnostic criteria.

All of the 6 included patients had symmetrical muscle weakness, which was diffuse in those with onset of symptoms up to 1 months of age, and either proximal or predominant in lower limbs .it was found that all of patients with SMA had homozygous deletions of exons 7 and 8 of the survival motor neuron 1 (SMN1) gene, that is one of the candidate genes identified within 5q13. Fasciculations, atrophy and decreased DTR were frequent findings.Laboratory metabolic tests and all brain CT scans were normal. EMG and NCV findings all showed normal motor and SNCV and denervation of muscles of upper and lower extremities that were compatible with a diagnosis of spinal muscular atrophy. Our results confirm that SMN1 copy number analysis is an important parameter for identification of couples at risk for having a child affected with SMA and reduces unwarranted prenatal diagnosis for SMA. Molecular studies can replace conventional investigations for SMA and have made the option of prenatal diagnosis possible for couples at risk.

References

Author Information

M. S. Omran
Department of Pediatric Neurology, Amirkola Pediatric Hospital, Babol medical University and Health Services

A. G. Juibary
Department of Pediatric Neurology, Amirkola Pediatric Hospital, Babol medical University and Health Services