Molecular and Epidemiological Characterization of SMN Genes in Cuban Patients with Spinal Muscular Atrophy

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Introduction: Spinal Muscular Atrophy (AME) is a neuromuscular disease of autosomal recessive inheritance with variable expressivity, characterized by degeneration and loss of the anterior horn neurons of the spinal cord and brainstem, resulting in progressive symmetric muscle weakness. The main cause of this disease is due to homozygous mutations in the SMN1 gene.

Objective: The aim of the current study was to characterize molecularly the SMN genotypes in Cuban patients affected with Spinal Muscular Atrophy by PCR of Restriction Fragment Length Polymorphism (RFLP), establishing their association with molecular and biostatistical techniques, with gender, age and skin color. This study has two main lines of work: the genotypic characterization by the detection of homozygous deletions of SMN1 and SMN2 by PCR-RFLP and the establishment of the relationship with the clinical phenotype, demographic or ethnic characteristics using statistical tests.The proportion of patients by provinces, type of SMA, sex and skin color was calculated.

Results: 74% of the patients presented homozygous deletion of SMN1 and 2, 56% had deletion of SMN2. In this study, cases with SMA were confirmed by molecular study, finding that the clinical and electrophysiological characteristics coincided with the data reported in the medical literature. 74% of the patients who fulfilled the clinical and electrophysiological criteria presented homozygous deletion of exon 7 of the SMN1 gene, which represents the mutation most frequently observed in patients with SMA. The distribution showed that the highest percentage of patients are concentrated in the western provinces of Cuba, especially in Havana, where the greatest number of specialized medical services are found where AME patients are treated and the diagnosis is made.

Conclusions: The results of this research seek to improve the genetic counseling of individuals carrying mutant SMN genes in order to improve quality of life by optimizing the diagnosis, prognosis and clinical management of these patients.

Key words: Spinal Muscular Atrophy, neuromuscular disease, genotypic characterization