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High proportion of mannosidosis and fucosidosis among lysosomal storage diseases in Cuba

C. Menéndez-Sainz[1], A. González-Quevedo[1], S. González-García[1], M. Peña-Sánchez[1] and R. Giugliani[2]

[1]Neurobiology Department, Institute of Neurology and Neurosurgery, Havana, Cuba
[2]Serviço de Genética Médica, Hospital das Clínicas de Porto Alegre, Departamento de Genética,
Instituto Nacional de Genética Médica Populacional,
Universidade Federal do Rio Grande do Sul, Porto Alegre, RS, Brasil

Corresponding author: C. Menéndez-Sainz
E-mail: cary@infomed.sld.cu

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ABSTRACT. Although lysosomal storage disorders (LSDs) are considered individually rare, as a group they present a non-negligible frequency. Few studies have been made of populational occurrence of LSDs; they have been conducted predominantly on Caucasian populations. We studied the occurrence of LSDs in Cuba. Data from individuals who had been referred to the Institute of Neurology and Neurosurgery in Havana from hospitals all over the country between January 1990 and December 2005 were analyzed. This institute was the only laboratory to provide enzyme-based diagnostic testing for 19 LSDs in Cuba during this period. Occurrence rates were calculated by dividing the number of postnatal diagnoses by the number of births during the study period. The combined occurrence of LSDs in Cuba was 5.6 per 100,000, lower than that reported in other studies conducted on Caucasian populations. The most frequent individual LSDs were: mucopolysaccharidosis type I (1.01 per 100,000) and,

surprisingly, alpha-mannosidosis (0.72 per 100,000) and fucosidosis (0.62 per 100,000). These findings may be related to specific genetic characteristics and admixture of the Cuban population. This is the first comprehensive study of the occurrence of LSDs in Cuba. We conclude that the epidemiology of these diseases can vary regionally, and we stress the need for similar surveys in other Latin American countries.

Key words: Inborn errors of metabolism; Lipidoses; Sphingolipidoses; Lysosomal storage diseases; Mucopolysaccharidosis

