
Miranda M¹, Larrinaga L¹, Acosta T¹, Charón DM¹, García Y², Valdes Y¹

(1) National Center for Medical Genetics, Havana-Cuba. misleidy@cngen.sld.cu (2) Provincial Center of Medical Genetics, Villa Clara-Cuba

**INTRODUCTION:** Mucolipidosis II / III (ML) are lysosomal storage diseases with an autosomal recessive inheritance pattern, caused by the deficiency of the Manose-6-phosphate marker responsible for generating the recognition signal, causing defects in processing and localization of acid hydrolases. N-acetylglucosamine-1-phosphotransferase is the deficient enzyme in ML, whose function is the transport of enzymes involved in the degradation of substrates within the lysosome. **OBJECTIVES:** To characterize the biochemical diagnosis of ML in Cuba in the last 5 years. **MATERIALS AND METHODS:** Six patients with clinical suspicion of ML were analyzed, with their relatives and healthy controls, for a total of 20 samples. The biological sample studied was blood serum. The specific enzymatic activity (AEE) was determined by fluorimetric method in the Shimadzu RF-5301pc equipment at a wavelength (λ) of emission 448 nm and of excitation 365 nm and spectrophotometric in the VS-850 equipment at 516 nm. **RESULTS:** Two patients diagnosed with ML II / III were diagnosed. The first patient, male, of 2 years of age, from the province of Santiago de Cuba, with a family history of this disorder and the second patient, female sex, 5 years old, from the province of Havana, both showed clinical manifestations similar to what is reported in this type of pathology. There was a considerable increase in AEE in the serum of the first patient and a decrease in AEE inside the lysosome in the second patient, in relation to the controls used. The specific enzymatic activity of patient 1 was used as a diagnostic criterion with respect to healthy control, exceeding its value more than 5 times, while in the case of patient 2 the value of the relative enzymatic activity was obtained from the comparison on the healthy control, assuming positive results less than 30%. **CONCLUSIONS:** In the last five years in Cuba of a total of 6 patients who were studied with clinical suspicion of Mucolipidosis II / III, only 2 were positive, which shows the low incidence of this disorder in our country. Key Words: Lysosomal storage disease, Mucolipidosis II / III, Enzymatic diagnosis.