P-067 - POMPE DISEASE IN CUBA. REPORT OF TWO CASES IN A FAMILY

Larrinaga L, Miranda M, Charón DM, Acosta T, Valdes Y, Llamos S

(1) Centro Nacional de Genética Médica, La Habana-Cuba. (2) Centro Provincial de Genética Médica de Guantánamo, Guantánamo-Cuba. emma@cngen.sld.cu La Habana-Cuba

INTRODUCTION: In the Inborn Errors of Metabolism are the Lysosomal Storage Diseases and one of them is the Pompe Disease that causes a progressive metabolic neuromuscular disorder due to the deficiency of the acid α-glucosidase (GAA) enzyme. This disease in its onset in childhood is characterized by cardiomyopathy, muscle weakness, respiratory disorders and early death. In this report we show two non-twin sisters who were born in different years with signs and symptoms suggestive of a disease presenting themselves in the first year of life.

OBJECTIVE: Perform the biochemical diagnosis of the Specific Enzymatic Activity (AEE) acid α-glucosidase and the Activity Relative Enzymatic (AER) of the two sisters.

MATERIALS AND METHODS: A biochemical study was carried out on the two sisters to detect enzymatic deficiency of GAA. In both cases the two patients were studied, a healthy control and the mother, the father was not studied. The biological sample used was heparinized whole blood, to which the leukocytes were extracted to perform total protein determination and subsequently determination of EEE (nmol / mgprot / h) and AER (%), both enzymatic determinations were performed by a fluorometric method, using 4-methylumbelliferone as a fluorogen. Fluorescence was measured on a Shimadzu RF-5301PC spectrofluorimeter, at an excitation wavelength of 365nm and emission of 448nm. The criterion used for the positive diagnosis is patient / control AER ≤ 30%.

RESULTS: The results obtained in 2014 for the first daughter were 12.36 nmol / mgprot / h and 30% and the mother 23 nmol / mgprot / h and 46%. The second girl born and studied in the year 2019 showed an EEE and AER respectively of 0.52 nmol / mgprot / h and 3% and the mother 27 nmol / mgprot / h and 50%. It was shown that both sisters are descendants of heterozygous parents because this is an autosomal recessive disease.

CONCLUSION: The diagnosis of Pompe disease was confirmed in both sisters.