P-077 - GAUCHER'S DISEASE PATIENTS PROFILE IN A REFERENCE SERVICE OF THE STATE OF AMAZONAS - BRAZIL

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INTRODUCTION: Gaucher’s disease is an inborn error of metabolism due to atypical lysosomal deposition (being the most common in this category), it is an autosomal recessive genetic disorder (chromosome 1p21) that compromises cellular functioning by accumulating glucocerebrosides, especially in macrophages. It is estimated to affect 1-9/100,000hab, manifesting itself under three clinical spectra, classified according to visceral (hepatosplenomegaly), hematologic (anemia and thrombocytopenia), bone (osteopenia and osteonecrosis), and nervous involvement and age of onset of symptoms, directly implicating in treatment and mortality rates. The diagnosis is guided by the clinical presentation, based on the activity levels of the malfunctioning enzyme – acid beta-glycosidase (ABG), and the detection of mutations, being the treatment done through enzyme replacement therapy (ERT) or inhibition of substrate synthesis.

OBJECTIVES: Describe epidemiological aspects of patients diagnosed with Gaucher’s disease in Amazonas.

MATERIALS AND METHODS: This is a descriptive observational study carried out in the public health system of the state.

RESULTS: At the moment, 16 patients are being treated, 8 of which are female and 8 are male, with a mean age of 16 years (ranging from 1 to 35 years) diagnosed at 10 years old (ranging from 2 to 34 years). All patients are treated with ERT (imiglucerase and alpha-taliglycerase), classified into types I and III of the disease and are, for the most part, below the expected rate of development and body mass.

CONCLUSIONS: and discussion: The registration of data like these is important since Brazil is the third in world incidence, therefore, it is essential to have knowledge about the disease and recognize it before the patient, once the therapeutic approach and diagnosis are able to predict and even avoid possible complications and repercussions.