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Niemann-Pick Disease: An Approach for Diagnosis in Adulthood

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Abstract

Niemann-Pick (NP) disease is a rare, autosomal recessive disorder characterized by visceromegaly and neurological alterations due to the excessive storage of lipids, sphingomyelin, and cholesterol. It commonly affects the child population, and only 6% of it occurs in the adult population. Type A is classified as the acute form, type B is the latest and with the best prognosis, and type C is characterized by neurological alteration. The diagnosis is based on enzymatic tests and genetic sequencing, with the latter being the diagnostic confirmation test. No specific treatment exists for this entity, although some patients with NPC type C may benefit from pharmacological treatment with miglustat.

The objective of this paper is to describe the clinical characteristics of a grown patient with Niemann-Pick diagnosis type B.

This article reports the case of a 55-year-old adult patient with a three-year clinical history consisting of splenomegaly and hematological disorders, without neurological symptoms ruling out frequent pathologies. Type B NP disease is diagnosed by a mutation in the sphingomyelin phosphodiesterase 1 (SMPD1) gene. The patient was receiving multidisciplinary support treatment.

Although NP disease is a rare disease according to the literature, it is important to consider this group of disorders as a differential diagnosis, when other more common pathologies have been ruled out in patients with isolated splenomegaly and thrombocytopenia

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Introduction

Lysosomal storage diseases are characterized by an enzymatic deficiency that involves the main function of lysosomes. Currently, there are 50 different kinds of heritage metabolic diseases [1]. Niemann-Pick (NP) disease, within this group, is an autosomal recessive disease. It is defined as the acid sphingomyelinase enzyme deficiency (SED), producing an alteration in the sphingomyelin and lipids deposits, thus causing a structural and functional change in the cell and viscera tissue [2]. The global prevalence is estimated between four and six cases per 1,000,000 of inhabitants [3] and is more frequent in the ascendant Jewish population. NP is diagnosed in 80% of cases under the age of 16 years, in 14% after 20 years of age, and extremely rare in older age, depending on the subtype [4-5].

Albert Niemann reported the first case in 1914, but in 1927, Ludwig Pick presented it as a different disease, marking its histological difference from the Gaucher disease [6]. The NP disease is classified based on organ involvement and enzymatic alteration. There are four sub-types (Types A, B, C, D), depending on the organ involved, age, and symptoms, with subtype B being the most frequent. With regard to enzymatic deficiency, in 1966, it was demonstrated that the acid sphingomyelinase enzyme deficiency (SED) in NP types A and B, but not in NP types C and D. Due to the above-mentioned, the four types of NP disease may be classified into two categories regarding the enzymatic deficiency: Type I is characterized by low acid sphingomyelinase levels seen in NP types A and B. In type II, a defect in low-density lipoprotein (LDL) transportation is seen in NP types C and D. The clinical presentation is related to visceromegaly and central nervous system involvement [2]. Diagnosis is based on clinical assessment and diagnostic tests, such as enzymatic quantification, fibroblast culture (Type C), and molecular biology techniques for identification (NPC1, NPC2, and SMPD-1 gene mutation identification). Later, a more specific genetic test should be made in order to obtain a specific diagnosis [7]. Treatment typically targets general symptom management: nevertheless, when there are neuropsychological disturbances, miglustat indication has shown a tendency toward delaying the neuronal degeneration in NP disease type C [2,8-9].

In Colombia, an orphan disease is defined as “A disease chronically debilitating, grave, life-threatening, and include rare diseases such as ultra-orphan diseases and forgotten diseases.” These diseases have more than