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Case Report

The Bone Marrow Cytological Diagnosis of Gaucher's Disease

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Abstract: Gaucher's disease is a rare lysosomal storage disorder with autosomal recessive inheritance caused by an enzymatic deficiency in glucocerebrosidases. The identification of Gaucher cells in cytology is a key marker but must be complemented by enzymatic and genetic analyses for a definitive diagnosis.

Keywords: Gaucher's Disease, Cytology, Gaucher Cells.

INTRODUCTION

Gaucher disease (GD) is a rare lysosomal storage disorder with autosomal recessive inheritance caused by an enzymatic deficiency of glucocerebrosidase. It is characterized by the accumulation of glucosylceramide in the lysosomes of macrophages, which take on a characteristic appearance and are called Gaucher cells, responsible for hematological, visceral, bone, and neurological impairments. The diagnosis of GD is made by measuring leukocyte glucocerebrosidase levels and is frequently suspected on the myelogram by the identification of Gaucher cells. Through this work, we demonstrate the crucial importance of cytological examination in the diagnostic orientation of Gaucher disease.

OBSERVATION

We report the case of a one-and-a-half-year-old infant, born to non-consanguineous parents, with no particular pathological history, who presented for a consultation due to a deterioration in general condition, hepatosplenomegaly without neurological involvement. The complete blood count showed pancytopenia. The bone marrow examination revealed the presence of a very large number of large macrophages, with monolobed nuclei with dense chromatin and cytoplasms that were slightly basophilic, having a 'crumpled paper' appearance, which are characteristic Gaucher cells indicative of Gaucher's disease (Figures 1 and 2).

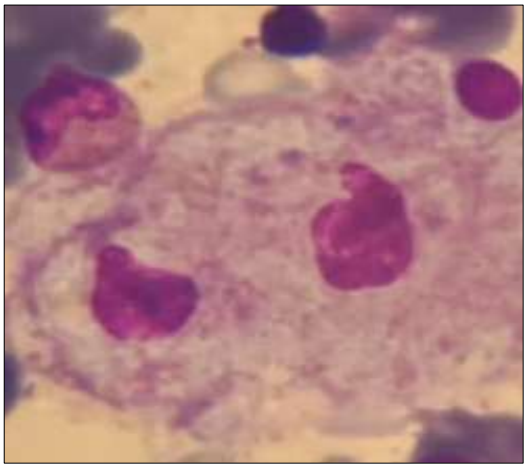

Figure 1: Gaucher cells in the bone marrow smear

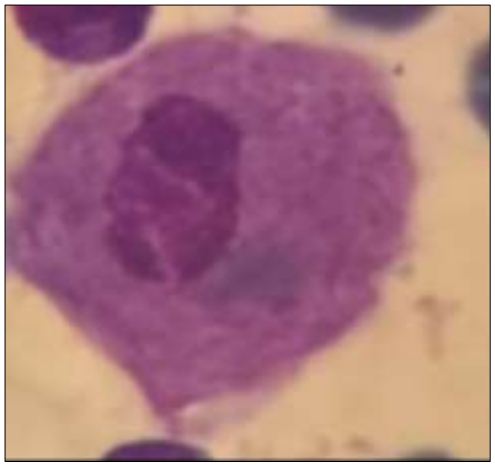

Figure 2: Gaucher cells in the myelogram

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DISCUSSION

Gaucher's disease is the most common of the storage disorders; it is caused by mutations in the GBA1 gene leading to a decrease in glucocerebrosidase activity. Gaucher cells infiltrate the hepatic and splenic tissue, the bone marrow, but also other organs, resulting in organomegaly and bone fragility.

The clinical context has great diagnostic value; thus, Gaucher's disease is a lysosomal lipid storage disorder of autosomal recessive inheritance. There are three types (1, 2, 3), with variable visceral, neurological, or hematological involvement. The clinical presentation of Gaucher's disease is heterogeneous, ranging from asymptomatic forms to lethal forms. Type 1, the most common, has a very variable phenotypic expression leading to cytopenias, splenomegaly which is one of the main clinical signs, often accompanied by hepatomegaly and bone involvement [1]. Types 2 and 3 are much rarer forms with severe neurological involvement and early death in type 2. Due to the usual modes of presentation of type 1 Gaucher disease, a myelogram is frequently performed in the face of cytopenia and splenomegaly; it helps guide the diagnosis by highlighting Gaucher cells. The diagnosis must be confirmed by measuring glucocerebrosidase activity in leukocytes, mononuclear cells, or fibroblasts [2, 3].

The cytological diagnosis of Gaucher's disease relies on the identification of characteristic cells called Gaucher cells in tissue samples, usually obtained through sternal puncture or bone marrow biopsy [3].

The cytological characteristics of Gaucher cells: morphologically, these are large cells (20–100 µm) of macrophage origin. The cytoplasm has a striated or "wrinkled paper" appearance (due to the accumulation of undegraded glucocerebrosides in the lysosomes). Eosinophilic or grayish on standard stains (May-Grünwald-Giemsa). The nucleus is eccentric, sometimes multiple, with fine chromatin. With the presence of fibrillary lysosomal inclusions visible under electron microscopy [4].

The techniques used:

- ✓ Myelogram (bone marrow puncture): First-choice examination to visualize Gaucher cells.
- ✓ Staining: May-Grünwald-Giemsa, PAS (positive), or acid phosphatase staining (positive reaction).
- ✓ Electron microscopy: Confirms lysosomal inclusions in "tubules" or pseudo-parallel structures.Le diagnostic cytologique oriente, mais nécessite une confirmation biochimique ou génétique.

The differential diagnosis is made with pseudo-Gaucher cells (observed in certain leukemias or blood disorders), with other lysosomal storage diseases such as Niemann-Pick disease (foamy cells, vacuolated cytoplasm), and Tay-Sachs disease (accumulation of GM2-ganglioside).

The diagnostic confirmation is made through enzyme dosage, showing a deficiency in glucocerebrosidase (acid β-glucosidase) in leukocytes or fibroblasts. Genetic confirmation involves searching for mutations in the GBA1 gene (chromosome 1q21). The analysis of the glucocerebrosidase gene (GBA1) located on chromosome 1 must then be carried out to identify the pathogenic variants of both alleles [5].

CONCLUSION

Gaucher's disease is a genetic disease that is likely underdiagnosed. The definitive diagnosis relies on demonstrating the enzymatic deficiency, which can be lengthy and difficult. In this context, cytology represents an important step in the diagnostic approach.

Conflict of Interest: The authors declare that they have no conflict of interest.

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