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Gaucher disease: About an observation

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Abstract

Introduction: Gaucher disease is an inherited lysosomal storage disorder caused by defective discomfort. The consequence is the deficiency or complete absence of an important enzyme, β-glucocerebrosidase, which controls specific metabolic processes in the body.Characterized by hematological, visceral and bone lesions and classified into 3 types. The main objective of our study is to describe the diagnostic peculiarity of our patient with Gaucher Disease.

Patient and Method: We report the observation of a child 6 years and 3 months old, history of prematurity, from a non-consanguineous marriage and a twin pregnancy. Since the age of 5 years, she has had arthralgia, bone pain and prolonged fever and whose examination found slightly discolored conjunctiva, arthralgia of the generals and a discreet splenomegaly.Laboratory assessment objectified hypochromic microcytic iron anemia, CRP and elevated ESR and imaging found enlargement of the distal metaphyses of both femurs on femur radio, slight homogeneous splenomegaly on the abdominal echo and cervico-thoraco-abdomino-pelvic CT scan found focus of condensation; Hepato-splenomegaly with a spleen containing fine calcifications; Isolated left ureteral dilation; Inhomogeneous bone pattern. B-Glucocerebrosidasa demonstrated low activity and genetic testing report was not detected for any pathogenic mutations.

Conclusion: If the symptoms of the disease are detected early and a correct diagnosis is made, organ damage can be prevented or mitigated by adequate treatment.

Keywords: Gaucher Disease; Glucocerebrosidase; Diagnostic Particularity; Organ damage

1. Introduction

Gaucher disease is an inherited lysosomal storage disorder caused by a defective gene. The consequence is the deficiency or complete absence of an important enzyme, β-glucocerebrosidase, which controls specific metabolic processes in the body.

Gaucher disease is a rare metabolic disease, it is estimated that fewer than 10,000 people worldwide are affected with only 1 in 40,000 patients with Gaucher disease.

Gaucher disease was very strictly classified into 3 types. This classification was based on the time of onset of the disease, the corresponding symptoms, the involvement of the nervous system, and the expectation

patients' lives. Today, a distinction is made between neuropathic and non-neuropathic forms of evolution.

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Gaucher cells most commonly accumulate in the spleen, liver, and bone marrow. However, the signs and symptoms of hematological, visceral and bone involvement. In Gaucher disease, inheritance is inherited autosomal recessive.

The suspicion of the diagnosis can be established on the basis of the individual symptoms as well as the laboratory results. Confirmation of the diagnosis is made by measuring the activity of the enzyme ß glucocerebrosidase in a blood sample. Gaucher disease has 2 different modes of treatment: enzyme replacement therapy or substrate reduction therapy.

2. Patient and Method

We report the observation of a child 6 years and 3 months old, with a history of prematurity, from a non-consanguineous marriage and a twin pregnancy.

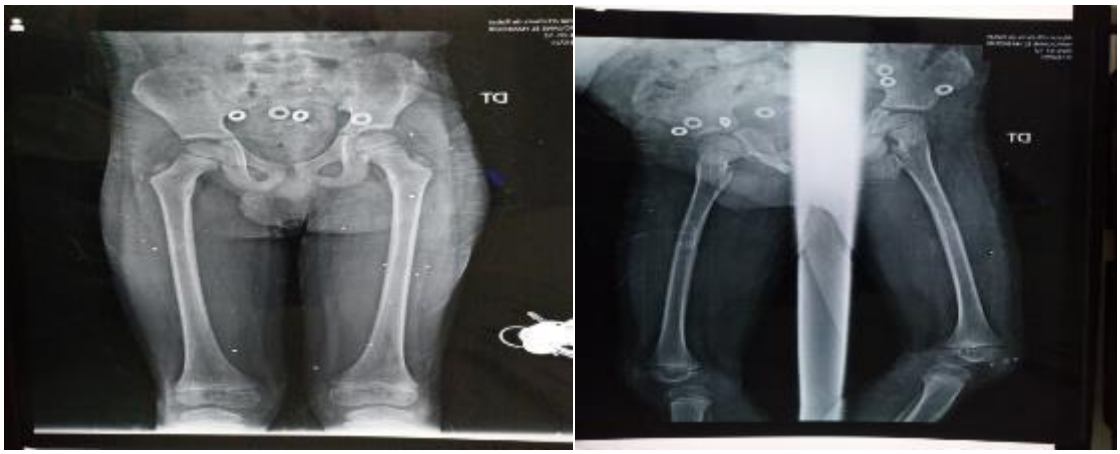

Figure 1 Femur Radio F+P: Enlargement of distal metaphyses of both femurs

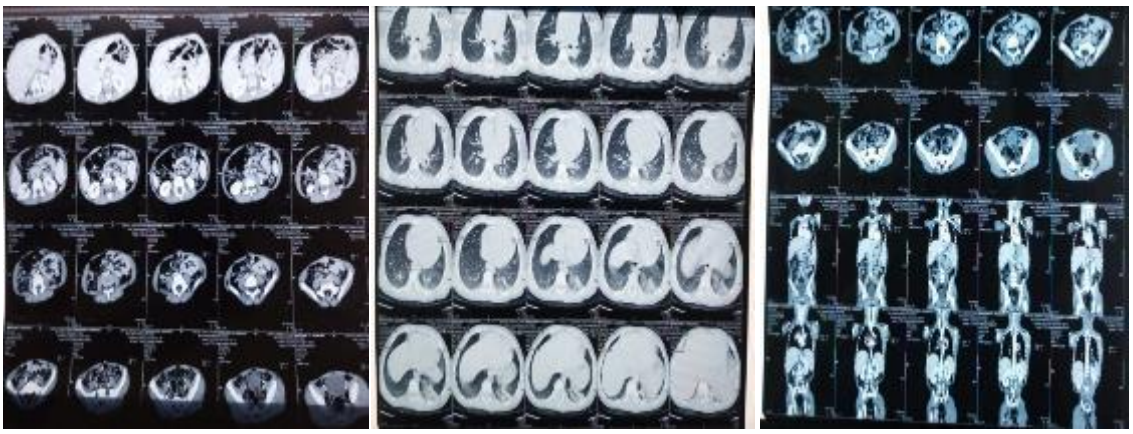

Figure 2 Cervico-thoraco-abdomino-pelvic scanner: Hepato-splenomegaly, Inhomogeneous bone frame

Present since the age of 5 years of arthralgia, bone pain and prolonged fever and whose examination found slightly discoloured conjunctivae, general arthralgia and a discreet splenomegaly. The biological balance objective hypochrome anemia microcytaires ferriprive, Transaminases and renal function are normal, high CRP and VS; Rheumatoid Factor, Ptsysio Balance, Cytobacteriological Examination of urine, Serologies Leishmaniasis and Aspergillosis are negatives. Myelogram with absence of blastic or malignant cells; Osteomedullar biopsy finds reactive hyperplastic marrow and absence of myelofibrosis. B-Glucocerebrosidasa activity showed low activity (below threshold value). The quantitative measurement of lyso-GL-1 was below the threshold value. Genetic Testing Report: No pathogen mutation was detected and no variant pathogen sequence was detected. The Imaginological assessment: Trans Thoracic Echo, Chest Radio, Dorso-lumbar spine Radio + Pelvis face are normal; Abdominal echo: Slight splenomegaly homogeneous; Femur Radio F+P: Enlargement of distal metaphyses of both femurs.(Figure 1) Cervico-Thoraco-Abdomino-Pelvic CT: Focal spot

of condensation; Hepato-splenomegaly with spleen containing fine calcifications; Isolated left ureteral dilation; Inhomogeneous bone frame (Figure 2)

3. Discussion

Gaucher's disease is a hereditary lysosomal overload disease caused by a defective gene. The consequence is the deficit or complete absence of an important enzyme, β-glucocerebroside, which controls specific metabolic processes in the body. (1,14)

In patients with Gaucher's disease, the function of the enzyme ß glucocerebroside is impaired, causing a disruption of metabolism.

The enzyme ß-glucocerebroside is one of the many enzymes normally found in what is called lysosomes.

If the body does not have enough ß-glucocerebroside, a sugar-containing fat called glucocerebroside accumulates in the lysosomes of so-called macrophages. These cells, typical of Gaucher disease, are also called Gaucher cells and can be detected in different organs. Due to the ever-increasing accumulation of glucocerebroside in lysosomes, Gaucher's disease is called lysosomal storage disease. With the accumulation of Gaucher cells in different organs, there are a variety of symptoms that increase in severity over time. Gaucher cells most often accumulate in the spleen, liver and bone marrow. (2,14)

They are much more rarely stored in other tissues, including the skin, eyes, lungs, heart and kidneys. In a limited number of cases, the nervous system may also be affected.

However, the main signs and symptoms are: risk of bleeding, anemia, fatigue, difficulty concentrating, decreased performance susceptibility to infections, increased spleen volume, increased volume of the time, bone pain, stunting, bone fractures risk significant bleeding, shortness of breath and nerve damage can cause eye movement disorders, seizures and other symptoms. (14)

In Gaucher disease, symptoms only occur if the affected individuals inherit the defective gene for ß glucocerebrosiidase from both parents. This is called autosomal recessive hereditary transmission, (14)

The suspicion of the diagnosis of Gaucher disease can be established on the basis of different symptoms as well as laboratory results. Confirmation of the diagnosis is made by measuring the activity of the enzyme ß glucocerebroside in a blood sample. In patients with Gaucher disease, enzymatic activity is greatly reduced.

In addition, genetic material analysis can be performed in patients to accurately determine the genetic defect.

Prenatal diagnosis is also possible. Specific tests are required to assess possible neurological involvement. (6,7,13,15)

Gaucher's disease was very strictly classified into 3 types. This classification was based on the time of onset of the disease, the corresponding symptoms, the involvement of the nervous system and hop of patients' lives.

This classification is increasingly neglected because there are transient forms that cannot be unequivocally attributed to a specific type. Today, it is possible to distinguish between neuropathic and neuropathic forms of evolution, that is, the occurrence or absence of nerve damage determines the assignment to one of the two main groups.

The non neuropathic form of Gaucher disease (formerly: type 1), a form of evolution without affecting the nervous system, occurs with a frequency of 90 to 95% and affects all age groups. The course of the disease can be very variable.

The neuropathic forms of Gaucher disease present the same symptoms as in the non neuropathic form, to which is added an involvement of the nervous system. Overall, these forms are very rare.

The form of acute progression with rapid deterioration (former designation: type 2) concerns young children. Symptoms neurological form of insidious chronic evolution (former classification: type 3) occur between early and late childhood and continue thereafter. (3,4,5)

Gaucher disease has 2 different treatment modes: Enzyme replacement therapy (TES) or substrate reduction therapy (TRS). Enzyme replacement therapy provides the body with the missing enzyme, glucocerebrosid, in the form of an infusion, every 14 days. The administered enzyme is artificially synthesized and mimics the action of endogenous glucocerebrosidasis. Enzyme replacement therapy can be used in all patients with type 1 and type 3 Gaucher disease.

Treatment by substrate reduction inhibits the synthesis of glucocerebroside, and is thus possible to prevent the accumulation of glucocerebroside in the body's phagocytes, which helps to mitigate or prevent damage to the organs, such as bone disorders or increased spleen and liver volume. (8, 9, 10, 11,12)

4. Conclusion

If symptoms of the disease are detected early and a correct diagnosis is made, organ damage can be prevented or mitigated by adequate treatment, leading to an improved quality of life.

Compliance with ethical standards

Disclosure of conflict of interest

No conflict of interest to be disclosed.

Statement of informed consent

Informed consent was obtained from all individual participants included in the study.

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