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Case Report

Gaucher disease discovered incidentally after splenectomy

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Abstract

*Gaucher disease is a genetically rare overload disease caused by a deficiency in a lysosomal enzyme beta-glucocerebrosidase. However, the discovery of Gaucher disease after splenectomy is rarely described in the literature. We report a clinical, radiological and biological observation of a man with splenomegaly with compression of neighbouring organs at the University- Hospital Center of Sidi Bel Abbes in ALGERIA, splenectomized for diagnosis and whose pathological examination has helped to guide the diagnosis and confirm by the assay of the enzymatic activity of glucocerebrosidase. This unexpected diagnosis of Gaucher disease pos*es a diagnostic problem when the clinical-radiological context is not concordant.

Keywords: *Gaucher disease, splenectomy, Gaucher Cell, Histology*
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Introduction:

Gaucher disease is a rare overload genetic pathology with recessive autosomal transmission, characterized by a deficiency in a lysosomal enzyme beta-glucocerebrosidase (1,2). This disease is characterized by glucosylceramide deposits in the splenic, hepatic and bone marrow cells where it results in hepatosplenomegaly sometimes associated with bone involvement and in some rare forms to neurological involvement (3). Non-neurological forms (type 1) are more frequent than neurological forms (types 2 and 3) (4). We report an observation of Gaucher disease diagnosed after splenectomy at the University- Hospital Center of Sidi Bel Abbes in ALGERIA.

Observation

This was a 40-year-old man from Sidi Bel Abbes city (located 400 km west of the capital Algiers) with normal past medical history, who complained of an easy fatigue lasting 3 months, with pain that had been evolving for two weeks in the left hypochondrium radiating to the left flank and back. On clinical examination, the patient was afebrile with Blood pressure of 110/75mmHg. On abdominal palpation there was a huge splenomegaly occupying almost the entire abdominal cavity. The abdomino-pelvic computed tomography scan showed splenomegaly of 30x15 cm with small nodules of 5mm disseminated throughout the spleen. There was no hepatomegaly and the neighbouring organs was compressed by the huge spleen. The blood count formula was normal (a hemoglobin level of 14.2 g/dL, a hematocrite value of 42%, a white blood cell count of 7000/mm3 and red blood cell count of 4900000/mm3) with a slight thrombocytopenia at 130 000/mm3. In this clinical and radiological context, especially the compression of neighbouring organs, a splenectomy was performed. The surgical specimen weighed 3500 gram and measured 28x15x8 cm, with a reddish-brown colour and rounded contours (figure 1), and showed numerous nodules measuring 05 mm without signs of necrosis on the cut -section (figure 2).

Figure 1: splenectomy piece fixed in 10% buffered formalin, measured 28x15x8 cm

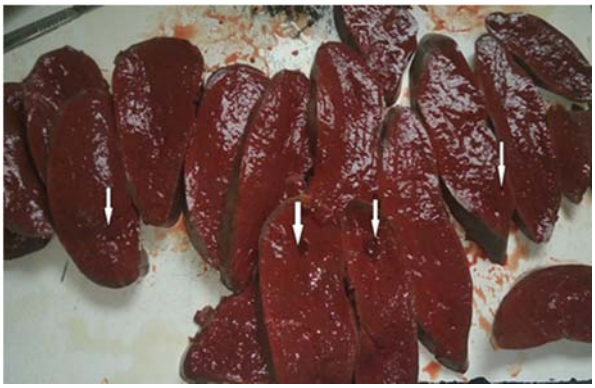

Figure 2: cut-section showed numerous nodules mesuring 5 mm without signs of necrosis

Histologically, the splenic parenchyma was interspersed with numerous macrophagic nodules separated by a few fibrous septa punctuated by a lympho-plasmocytic inflammatory infiltrate, these macrophages are large cells, with fibrillar eosinophilic cytoplasm and with eccentric nucleus of crumpled appearance very characteristic of Gaucher cells. The immunohistochemical study by CD 68 antibody showed diffuse, homogeneous, intense cytoplasmic and membrane labelling of macrophages (Gaucher cells) (figure 3A and 3B).

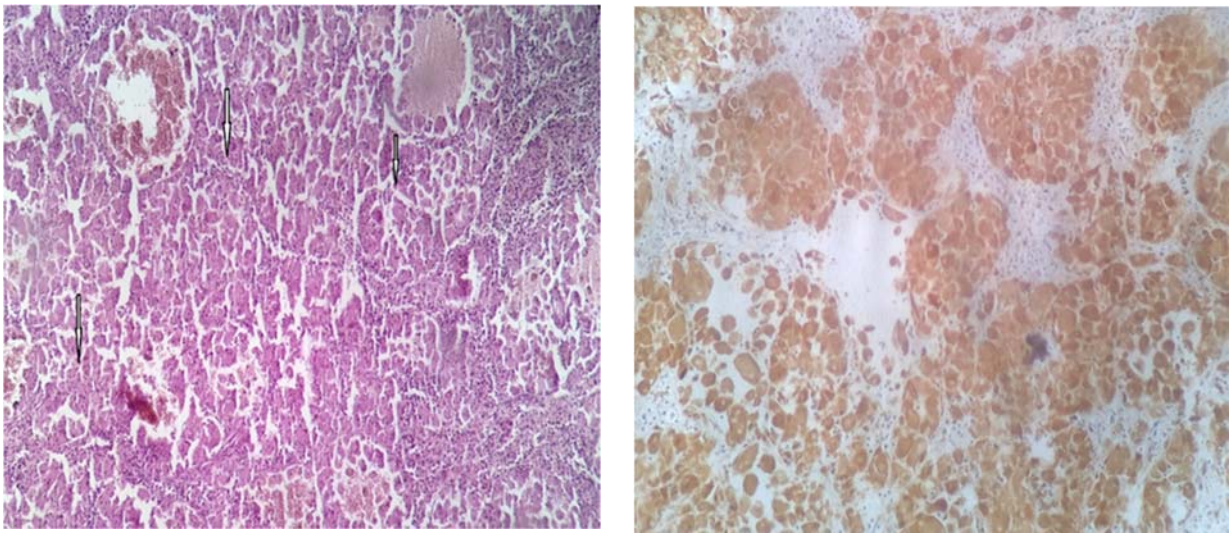

Figure 3:(A) microscopic showing numerous macrophagic nodules separated by a few fibrous septa punctuated by a lympho-plasmocytic inflammatory infiltrate, these macrophages are large cells, with fibrillar eosinophilic cytoplasm and with eccentric nucleus of crumpled appearance very characteristic of Gaucher cells (hematoxylin & eosin, X200), (**B**) microscopic showing diffuse brown, homogeneous, intense cytoplasmic and membrane labeling of macrophages (Gaucher cells) by the antibody CD 68 in the immunohistochemistry study, X200.

In view of this discovery, the diagnosis of Gaucher disease was strongly suspected and the enzymatic analysis found an increase in the enzymatic activity of glucocerebrosidase, which confirmed the diagnosis. A scintigraphy bone scan (99mTc) was performed and found no location and the neurological examination was normal.

Discussion

Gaucher disease was first described by Dr Gaucher in 1882 and its prevalence is around 1/100 000 in the general population (5). However, the diagnosis of Gaucher disease after splenectomy, has been very rarely reported in the literature. Only one observation has been reported in a series of 04 cases by *Adas et al* (6). The age at the time of diagnosis is extremely variable from 0 to 90 years, usually before 10 years, our case is 40 years old. Three types of Gaucher disease are described: type 1, defined by the absence of neurological involvement, representing 94%. Type 2, characterised by early neurological involvement, is the rarest and most severe form 1%. Type 3, characterised by later neurological involvement and a more progressive course, accounts for 5% of the cases. Our patient was classified as type 1 because there was only splenomegaly, no hepatomegaly, no neurological involvement, and no bone involvement. Our observation underlines the importance of anatomopathological examination in the diagnosis by showing nodules of macrophagic cells with a crumpled appearance characteristic of Gaucher cells (8). In this case, the determination of leukocyte beta-glucocerebrosidase enzyme activity confirmed the diagnosis.

Treatment of Gaucher disease is primarily medical therapy with intravenous enzyme replacement therapy, splenectomy is only considered in case of haematological complications: hypersplenism, haemorrhagic syndrome (9). In our patient, splenectomy was performed for diagnostic purposes. The evolution of Gaucher disease is generally favourable with a mortality rate of around 2% (10) and the evolution in our patient was favourable after splenectomy.

Conclusion

Although the diagnosis of Gaucher disease after splenectomy is a difficult one to make when the clinical and radiological features are discordant, it should always be sought, while emphasising the importance of pathological examination in establishing the diagnosis.

Conflict of interest

The authors declare no conflict of interest

Author contribution

All the authors contributed equally to the manuscript. They approved the final and revised version of the manuscript.

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