Huge Subcapsular Splenic Hematoma in a Patient with Gaucher Disease

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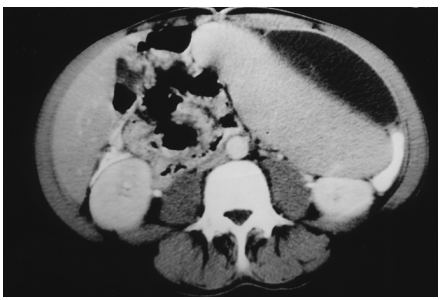
Gaucher disease is usually associated with splenomegaly. We describe here a 45-year-old man, recently diagnosed with Gaucher disease, who presented with massive splenomegaly that retrospectively was found to be caused by a huge subcapsular splenic hematoma. Follow-up included ultrasound and computerized tomography scan for 9 months when disappearance of the hematoma was noted.

Case Description

In May 1998, a 45-year-old Ashkenazi man experienced severe left flank pain unrelated to any trauma, although he remembers having lifted a rather large stone before onset of the abdominal pain. Physical examination in another medical center at that time revealed splenomegaly and generalized lymphadenopathy. Complete blood count showed hemoglobin 11.5 g/L, white blood cell count 2,800/mm^3, and platelet count 48,000/mm^3. Bone marrow aspiration revealed Gaucher cells. Treatment with enzyme replacement therapy was begun in September 1998.



Subcapsular splenic fluid collection on CT

Two months later the patient appeared in our Gaucher clinic for further consultation. Physical examination revealed hepatomegaly and massive splenomegaly. As part of the routine evaluation, abdominal ultrasound was performed, which showed splenomegaly (18.6x18x10 cm^3) and a large hypoechoic subcapsular collection (12x5x13 cm^3) compatible with an old hematoma [Figure]. In addition, hepatomegaly and the "Erlenmeyer flask" deformity of the distal femur were noted. The patient was treated conservatively with bed rest. CT scan a month later showed shrinkage of the hematoma. Abdominal ultrasound performed 7 months later, in July 1999, showed complete resorption of the hematoma.

Comment

Gaucher disease, the most prevalent sphingolipid storage disorder, is caused by an autosomal recessive defect of the lysosomal enzyme, β-glucocerebrosidase, and results in accumulation of glucocerebroside in

the cells of the monocyte-macrophage system. The diagnosis of Gaucher disease is based on a biochemical assay of enzyme activity and confirmed by polymerase chain reaction-based DNA mutation analysis. Enzyme replacement therapy has proven to be both safe and effective in reducing the organomegaly and improving the hematological parameters in symptomatic Gaucher patients [1].

Subcapsular hematoma has rarely been described in Gaucher disease, even among those with massive splenomegaly and severe hypersplenism or thrombocytopenia. Hancock [2] described a subcapsular splenic hematoma in a 28-year-old healthy patient after strenuous exercise on the cricket field. This case was reported in 1971, before the advent of CT scans and sophisticated sonography, and hence the diagnosis of the hematoma was intraoperative. During surgery the hematoma was aspirated, the spleen removed, and Gaucher cells were noted in the pathological specimen.

In more recent studies evaluating the spleen in patients with Gaucher disease, only one of 52 patients with ultrasound examination [3] had a nearly complete infarction with a subcapsular hematoma that subsequently required splenectomy. Of 46 patients undergoing magnetic resonance imaging [4], only 4 patients had small subcapsular fluid collections with subcapsular splenic infarcts.

The case presented here is the first among our group of more than 375 Gaucher patients. The etiology in this patient may have been viral (perhaps subclinical infection with Epstein-Barr virus), which aggravated concurrent hypersplenism and thrombocytopenia; or as in Hancock's case may be due to physical exertion without trauma.

The hematoma was clearly demonstrated on abdominal ultrasound. We routinely use ultrasonography in all our Gaucher patients on initial evaluation and at follow-up since it is noninvasive, inexpensive, and does not involve repeated exposure to radiation. In this case, an unusual finding that was missed on physical examination and early evaluation was accurately discovered by ultrasonography and its course was documented.

References

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