Wandering Spleen in a Young Girl with Gaucher Disease

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Wandering spleen is a rare entity in which free-floating splenic tissue is attached by a vascular pedicle without peritoneal attachments. Malformations, laxity, or absence of appropriate ligaments may lead to ptosis and malposition of the spleen in the abdomen or pelvis. This abnormally positioned spleen – referred to as a wandering, pelvic, or floating spleen – is prone to intermittent twisting on its vascular pedicle that causes characteristic pain as well as ischemia and splenic infarcts.

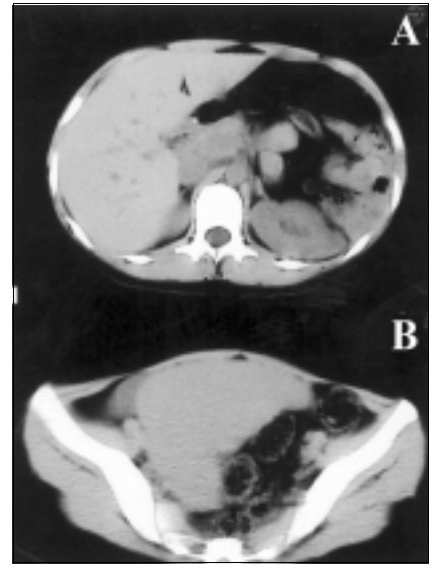
We describe a girl with Gaucher disease whose presenting symptoms were abdominal pain and a large abdominal mass that was diagnosed as a wandering spleen. Given that splenectomy is only rarely recommended in Gaucher disease, the treatment of choice for this patient was initially enzyme replacement therapy in an attempt to reduce the size of the spleen.

Case Description

In June 1995, a 12 year old Ashkenazi girl experienced epigastric and left-sided abdominal pain. A complete blood count showed hemoglobin 10.1 g/L, white blood cell count 3,200/mm^3 and platelet count 64,000/mm^3. Ultrasound revealed an enlarged homogenous spleen in an abnormal location, and hepatomegaly. Bone marrow aspiration taken in another medical center demonstrated Gaucher cells.

A month later the patient was seen in our referral clinic for Gaucher disease. The spleen was palpated as a large nontender mass measuring approximately 20 x 16 cm in the middle-to-left side of the abdomen and extending into the pubis; Traube's triangle was free. The liver was palpable 3.5 cm below the costal margin; abdominal circumference measured 66.5 cm. Computerized tomography documented the position of the spleen [Figure]; the volumetric measurement of the spleen was 1,263 cm^3 and of the liver 1,256 cm^3.

In December 1995 low dose enzyme replacement therapy (30 units/kg/month administered biweekly) was started in the hope that it would reduce the massive splenomegaly [1]. The patient received enzyme replacement therapy for 4 years but intermittently continued to experience episodes of abdominal pain. Physical examination at the most recent follow-up revealed appropriate linear



CT images taken in November 1995 at the level of the kidneys where the head of the pancreas should normally appear **[A]**, and at the level of the iliac bones where the body and tail of the spleen are actually seen **[B]**.

growth (75th percentile for age) and a spleen measuring 12 x 15 cm with an abdominal circumference of 60.5 cm. Volumetric measurement by CT of the spleen was 1,367cm^3 and of the liver 1,372 cm^3.

Comment

A wandering spleen is rarely diagnosed, especially in children. The incidence is highest in adults aged 20–40 years, with 70–80% of the cases being women of reproductive age. Children account for only one-third of the cases, 30% of whom are under 10 years of age [2]. Although there is no standard radiographic test for diagnosis, duplex ultrasonography and CT are most commonly employed.

The definitive treatment for wandering spleen is surgery since non-operative treatment is associated with a complication rate as high as 65% [3]. Complications include infarction, gangrene, splenic abscess, variceal hemorrhage and pancreatic necrosis [2] that may lead to an acute abdomen. Historically, splenectomy was the treatment of choice for wandering spleen. However, in view of the heightened appreciation of the importance of the spleen in the reticuloendothelial system, splenoplexy with splenic salvage is currently the procedure of choice for wandering spleen [3]. Acute cases of splenic torsion with consequent infarction still necessitate splenectomy.

Gaucher disease invariably results in splenomegaly, but this is the only case of wandering spleen among more than 400 patients with Gaucher disease in our clinic. As mentioned, splenectomy was traditionally recommended in Gaucher disease, however the advent of enzyme replacement therapy has markedly limited indications for this procedure to a few specific situations, such as failed enzyme therapy or the critical need for surgery in a patient with severe thrombocytopenia [4]. Although our patient has had no further episodes of splenic torsion, the choice of enzyme replacement therapy was not optimally effective since there was no significant reduction in splenomegaly over time. A therapeutic option for this patient may be splenoplexy, particularly since future fertility must be taken into account given that pregnancy per se may be a predisposing factor for wandering spleen [5]. Similarly, splenectomy cannot be ruled out in this particular instance if mechanical obstruction during pregnancy or repeat episodes of torsion occur.

Of interest is the fact that this patient has two siblings with the same genotype who are asymptomatic and whose respective spleen and liver are not palpable on physical examination and are only slightly enlarged by ultrasound examination. Although identical genotypes do not necessarily have identical phenotypic expression, this patient might have been asymptomatic had she not had a wandering spleen.

References

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