Iliopsoas Hematoma in Gaucher’s Disease

Jéréme Selton, Julien Perrin, Hélène Ropion, Manal Abdelfatah, Boualem Siouala, Lelia Pruna and Pierre Kaminsky

Abstract

Gaucher’s disease is an autosomal recessive inherited disease characterized by organomegaly, cytopenia and bone destruction. Clotting disorders and platelet dysfunctions are described. We report the case of a 22-year-old man who presented subacute groin pain due to spontaneous iliopsoas hematoma. Laboratory investigations found moderate thrombocytopenia, normal coagulation factor activities and unspecific platelet function test disturbances. His spleen was moderately enlarged and no significant bone lesions were found. Iliopsoas hematoma is a rare complication in Gaucher’s disease and should be included in the differential diagnosis of pain localized to the groin-hip area, which could rather evoke hip osteonecrosis in this context.

Key words: Gaucher disease, Iliopsoas hematoma, Platelet function, Coagulation disorder

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Introduction

Gaucher’s disease (GD1) is an inherited autosomal recessive disorder due to a deficiency in glucocerebrosidase, a lysosomal enzyme that catalyses the hydrolysis of the glycosphingolipid glucocerebroside to ceramide and glucose (1). Lysosomal storage occurs in reticuloendothelial cells and leads to hepatomegaly and splenomegaly, which causes hypersplenism and cytopenia. Bleeding is a frequent symptom in GD1. It is usually attributed to thrombocytopenia, although deficiencies of various coagulation factors and abnormal platelet functions have been reported (2-4). However, the skeletal manifestations are probably the most disabling aspect of the disease. Patients commonly experience bone pain, and some suffer bone crises (1). Radiological findings include Erlenmeyer flask deformity, bone marrow infiltration, osteopenia, osteosclerosis, fractures and osteonecrosis (1).

We report the case of a young patient with type I GD1 who presented with groin pains which revealed iliopsoas hematoma despite moderate thrombocytopenia and normal coagulation tests.

Case Report

A 22-year-old man presented with a sudden onset increasing pain in the left groin and hip radiating to the left anterior thigh. The patient was not on any medication and no history of trauma was given. GD1 was diagnosed at the age of 2 years, based on findings of moderate thrombocytopenia associated with moderately enlarged spleen. Gaucher’s cells were found in myelogram and low glucocerebrosidase levels were demonstrated in leucocytes. He was treated with low doses of imiglucerase (400 U), but the patient and his parents refused regular follow-up and posology adjustment with growth. At admission, physical examination revealed flexion deformity of the hip, and decreased quadriceps reflex. His spleen was palpable 5 cm below the costal margin. Diagnosis of hip osteonecrosis or vertebra fracture was first suggested, however the hip and spine radiographs were normal. Laboratory tests included the following: hemoglobin 14.5 g/dL, platelet count 70,000/μL, aPTT 39s/33s and PT 83%. At day 1, hip pain increased requiring morphine administration and the patient complained from thigh flexion weakness. Hemoglobin level decreased by 2 g/dL (Table 1). As a consequence, CT scan of hip and spine was performed. Bone

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Table 1.  Time Evolution of Clinical Symptoms, Radiological Findings and Hematological Investigations

<table>
<thead>
<tr>
<th></th>
<th>Admission</th>
<th>Day 1</th>
<th>Day 4</th>
<th>Day 45</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Clinical symptoms</strong></td>
<td>pain of left groin and hip</td>
<td>increasing pain requiring morphine administration</td>
<td>decreasing pain</td>
<td>discharged at day 8</td>
</tr>
<tr>
<td><strong>Imaging</strong></td>
<td>normal radiographs of hip and spine</td>
<td>CT: 12 cm iliopsoas hematoma</td>
<td>echography: 8 cm hematoma</td>
<td>CT: normal aspect of iliopsoas muscles</td>
</tr>
<tr>
<td><strong>Biology</strong></td>
<td>Hb: 14.5 g/dL, Platelets: 86,000/μL</td>
<td>Hb: 12.2 g/dL, Platelets: 70,000/μL</td>
<td>Hb: 12.2 g/dL, Platelets: 146,000/μL</td>
<td>Hb: 12.7 g/dL, Pl: 90,000/μL</td>
</tr>
<tr>
<td><strong>Coagulation tests</strong></td>
<td>PT: 86%, aPTT: 42s/34s</td>
<td>PT: 83%, aPTT: 39s/30s, TT: 16s/20s</td>
<td>factor XII: 69%, factor XI: 61%, factor IX: 78%, factor VIII: 220%</td>
<td>factor XII: 69%, factor XI: 61%, factor IX: 78%</td>
</tr>
</tbody>
</table>

Structures were normal but a 12-cm hematoma of the left iliac psoas muscle was found (Fig. 1). Due to increasing pain, the patient was then referred to our hospital on day 5 and since neurological symptoms were minor, no surgery was performed. In addition, the size of hematoma was found to be decreased using echography measurement (Table 1). Assessment of coagulation factor activity revealed factor XII at 69%, factor XI 61%, factor IX 78%, factor VIII 220%. Bleeding time was 9 minutes. Von Willebrand factor antigen was 215%, and plasma von Willebrand factor activity 188%. The platelet aggregation was normal in response to arachidonic acid, ristocetin and collagen, reduced in response to epinephrine (5 and 10 μM) and reduced in response to ADP 5 μM, but normal to ADP 10 μM. Pain was controlled using morphinic drug; his muscle strength recovered and the patient was discharged. Six weeks later, CT scan and MRI were performed in order to detect GD-related complications. They showed spontaneous dissolution of the hematoma (Fig. 1). Gaucher’s disease was confirmed by very low glucocerebrosidase levels at 1 nmol/h/mg protein (N: 10-30), and chitriosidase level was dramatically increased at 4,180 nmol/h/ml (N<60). Reduction in both T1 and T2 marrow signals was found in spine and heterogenous T1 and T2 signals were observed in femurs without bone deformity or cortical resorption (Fig. 2). MRI of the abdomen showed spleen enlargement at 16 cm.

**Discussion**

Iliopsoas hematoma usually occurs in patients undergoing acute or chronic anticoagulant therapy or suffering from acquired or inherited clotting disorders (5). It is noteworthy that several reports have referred to traumatic hematomas or to spontaneous hematoma without coagulation abnormality (5), or to muscular hematoma due to thrombocytopenia (6). Five previous GD1 patients have been previously reported to present spontaneous iliopsoas hematoma (7, 8). These reports are summarized in Table 2. Since such hematomas are rare (5), and taking into account the very low prevalence of GD1, this association is therefore more than merely coincidental.

Bleeding is a common feature in GD1 patients, not only owing to thrombocytopenia, but also because of abnormal clotting and of platelet dysfunction. This can lead to serious problems during pregnancy or surgery (2). In a series of 30 patients with GD1, prolonged aPTT and PT were found in 42% and 38% of patients, respectively. Low levels of coagulation factors XII, XI, VII, X, or V have been also reported in GD1 (2, 3). These abnormalities were linked to the presence and size of the spleen (3). Moreover, Gillis et al have reported abnormal platelet aggregation tests in 22% of GD1 patients (4). They concluded that platelet dysfunction is a relatively common cause of excessive bleeding in Gaucher’s disease patients.

Muscular hematoma typically occurs in patients with severe coagulation disorders, such as hemophilia or von Willebrand’s disease, or with anticoagulant agents, all conditions characterized by a significant increase in aPTT or PT due to inherited or acquired low coagulation factor levels in blood. Although the present patient exhibited decreased levels in several coagulation factors, the observed abnormalities seem to be sufficiently modest to not really explain the occurrence.
Figure 1. Pelvis tomodensitometry at admission (A, B, C, D). A: coronal reconstruction showing normal aspect of the hip. B: Axial slice showing enlargement of the left psoas muscle with an infiltrative aspect of perimuscular tissues (arrow). C: Axial slice demonstrating enlargement of both psoas and iliac muscles by a heterogeneous mass (arrows). D: coronal reconstruction showing the iliopsoas hematoma (arrows). E: CT scan performed 6 weeks later demonstrating normal aspect of both iliac and psoas muscles.

Figure 2. Left: abdominal and pelvic CT scan. Coronal reconstruction showing normal aspect of the liver and enlarged spleen with normal density. Middle and right: Coronal MRI section of the left femur. While the hip showed the normal aspect, femoral diaphysis showed T1-weighted hyposignal (middle) and a heterogeneous signal in fat-suppressed T2-weighted sequence, corresponding to moderate bone infiltration by Gaucher’s cells.
Table 2. Summary of the Clinical and Biological Findings in the Five Previous Reports of Iliopsoas Hematoma in Patients with Gaucher’s Disease

<table>
<thead>
<tr>
<th>Reference</th>
<th>gender; age (years)</th>
<th>Spleen, Liver, Bone</th>
<th>spontaneous bleeding</th>
<th>hemogram</th>
<th>Coagulation tests</th>
<th>ERT</th>
</tr>
</thead>
<tbody>
<tr>
<td>Flipo [7]</td>
<td>F: 20y SPX enlarged (16cm)</td>
<td>spine: diffuse hypoosignal in T2-weighted sequence</td>
<td>no</td>
<td>Hb: 11.0 g/L</td>
<td>Platelets: N</td>
<td>PT : 60% aPTT : 39.5s/30s FII : 70% FV : 57% FVII + FX : 70% FXI: 32% normal bleeding time</td>
</tr>
</tbody>
</table>

SPX: splenectomized. Hb: hemoglobin, coag: coagulation tests, ERT: enzyme replacement therapy, ?: data not available

of severe bleeding. It is noteworthy that no major abnormalities of coagulation were reported among the five previous reports of iliopsoas hematoma in GD1 patients (Table 2), even though details of coagulation tests were given only in one (7). Moreover, psoas hematoma is very uncommon in patients suffering from thrombocytopenia or from platelet dysfunction. Furthermore neither the present patient nor the five previously reported patients exhibited significant thrombocytopenia, platelet counts being always higher than 50,000/μL, nor severe platelet dysfunction. As a consequence, it is unlikely that decreased platelet count or platelet function could explain the occurrence of psoas hematoma. Moreover, several drugs or trauma can also promote muscular hematoma. In this way, Lesić et al reported strenuous exercise in three of their cases (8). However, in the present case report as in that of Flipo et al (7), no particularly intense exercise or drug intake could be found, leading to the diagnosis of “spontaneous” hematoma. The liability of the underlying moderate coagulation abnormalities remains a matter for discussion.

Interestingly, all 5 of the previously reported patients were young and suffered from severe GD1 with significant bone or visceral overload, and two of them underwent splenectomy (Table 2). All 5 presented with significant skeletal involvement or bone crisis. The present case is in contrast to these earlier reports, since only moderate splenomegaly and bone lesions were observed.

Physiopathology of iliopsoas hematoma thus remains unclarified. In clinical practice, although iliopsoas hematoma is a rare complication in GD1, it should be included in the differential diagnosis of pain localized to the groin-hip area, which could rather evoke, in this context, hip osteonecrosis or spine complications. Additionally, caution is required because the risk of bleeding of orthopedic surgery must be considered (4).

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References

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