Iliopsoas Hematoma in a Young Patient with Type I Gaucher Disease

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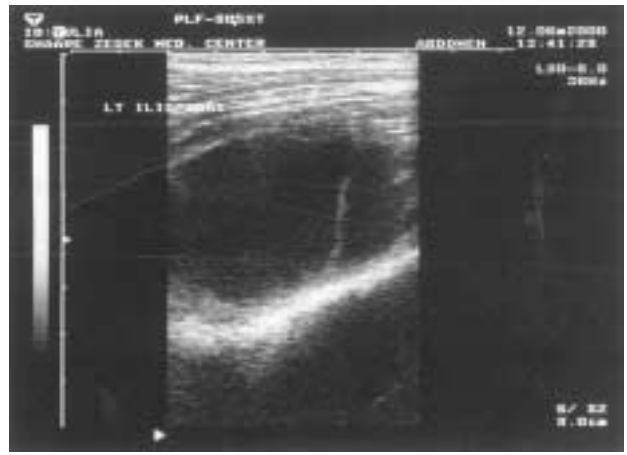
Gaucher disease, the most prevalent lysosomal storage disorder, is a result of a genetic defect in the enzyme β-glucocerebrosidase. The most common symptomatic presentation includes hepatosplenomegaly, as well as anemia and thrombocytopenia due to hypersplenism, and often, skeletal involvement [1]; however, spontaneous bleeding is extremely rare. The current case is a girl with severe skeletal involvement who presented with groin pain and inability to walk.

Patient Description

A 15 year old girl was diagnosed in Russia with Gaucher disease 6 years after undergoing splenectomy. Within a year of the splenectomy she experienced recurrent episodes of bone crises along with avascular necrosis of the left hip, bone deformities, and pathologic fractures in both tibias and both shoulders. Bleeding complicated consequent surgeries. Repeated courses of high dose steroids were routinely given. The most recent operation was an unsuccessful attempt to correct a 4 cm limb-length discrepancy. Due to the exquisite pain in the left groin area, the patient was immediately sent to Israel for treatment.

She presented to our clinic as a pale child of normal height for age (50th percentile), but underweight (3rd percentile). She had limitation in rotation and abduction of both hip joints, tenderness over the left hip joint, with slight limitation on flexion of both joints. The abdomen was tender in the left lower quadrant, and the liver was palpable 6 cm below the costal margin. There was a positive psoas sign on the left side. Tanner score was 2. Laboratory tests included: erythrocyte sedimentation rate 116, white blood cells 11,100/mm^3, hemoglobin 9.1 g/dl, mean cell volume 80.2, platelet count 724,000/mm^3, and abnormal liver tests. X-rays revealed severe osteoporosis, cervical gibbous formation, avascular necrosis of the left hip, shin splints, pathologic fractures in both tibias, and overt bowing of both humeri. Both heels had multiple fractures. Abdominal ultrasound showed hepatomegaly and cholelithiasis. A collection in the iliopsoas muscle was noted in the left groin [Figure].



Abdominal ultrasound showing collection in the iliopsoas muscle in the left groin.

Sequencing of the β-glucocerebrosidase gene revealed the genotype N370S/W381STOP; the latter mutation has not been described before.

Management

Ultrasound-guided draining of the collection in the iliopsoas muscle was performed immediately with evacuation of 75 ml of blood-stained opaque fluid. Drainage continued with concomitant antibiotics for 1 week. Within hours of drainage, the patient felt better and was able to walk; the sedimentation rate and white blood count decreased and cultures from drainage fluid and blood were negative.

Enzyme replacement therapy [2] was instituted during her hospitalization, and after 1 year the patient had gained weight and undergone a compensatory spurt in height along with sexual maturation. Anemia and the tendency to bleeding were normalized. There was no reversal of the skeletal pathologies. She returned to Russia and is doing well.

Comment

Iliopsoas hematoma presents as severe and persistent pain in the groin, radiating upward to the lumbar area or distally along the thigh. The hip is held in flexion and in external rotation because of spasm of the iliopsoas muscle. Any attempt to straighten the leg results in intensification of the pain. A palpable mass in the iliac fossa may be found. Pressure on the femoral nerve may lead to femoral palsy. Differential diagnosis includes iliopsoas bursitis, malignant infiltration, hemarthrosis of the hip joint, and iliopsoas abscess. Iliopsoas hematoma is most often seen in individuals with hemophilia [3] or requiring anticoagulation [4].

The diagnosis is made using imaging techniques, but sonography is generally preferred for demarcation. Therapeutic approaches include guided drainage [5], percutaneous decompression, or bed-rest, and/or specific treatment, e.g., anticoagulation withdrawal or factor replacement in hemophiliacs. In this patient, the initial diagnosis was challenging because symptoms included features of abscess (fever, malaise, elevated white blood cell count and sedimentation rate) as well as those of hematoma (exquisite pain and inability to straighten the leg).

Coagulation abnormalities in Gaucher disease are generally due to thrombocytopenia and/or poor platelet function, as well as concomitant coagulation factor deficiencies [5]. In our referral clinic with more than 475 patients, there has been only one case of a spontaneous hematoma (of the spleen). Thus, a bleeding etiology as seen in hemophiliacs and patients with anticoagulation is unexpected in Gaucher disease; however, this patient had a history of heavy bleeding after surgical interventions and hence may be comparable to classic cases of iliopsoas hematoma.

Of interest was the severity of the bone involvement in our patient, which affected virtually all long bones and the spine. The patient is a compound heterozygote for a common mild mutation, N370S, and a novel null mutation, W381STOP (c. 1259G>A/g.5291G>A), the latter creating a premature stop codon in exon 9. A combination of mild and null mutations is generally associated with severe, albeit non-neuropathic, type I Gaucher disease. It may be surmised, therefore, that the patient's initial presentation of a severe phenotype reflected this genotype, but that the natural course of her disease has been modified by palliative therapeutic procedures and enzyme replacement therapy.

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