Case report

Severe skeletal involvement in a patient with Gaucher’s disease

Hatice Nursun Özcan1, Murat Kara2, Özgür Kara3, Funda Pepedil3, and Levent Özçakar2

1 Department of Radiology, Ankara Numune Education and Training Hospital, Ankara, Turkey
2 Department of Physical Medicine and Rehabilitation, Hacettepe University Medical School, Ankara, Turkey
3 Department of Internal Medicine, Hacettepe University Medical School, Ankara, Turkey

Introduction

Gaucher’s disease (GD) is a rare but progressive lysosomal storage disorder caused by β-glucocerebrosidase deficiency. Accordingly, glucocerebroside accumulates in the reticuloendothelial system and various musculoskeletal complications (i.e., osteopenia, abnormal bone remodeling, delayed bone healing, pathological fracture, increased propensity for infection, and bleeding ensue due to involvement of the bone marrow).1 Likewise, in this report, we present a challenging case of GD that was accompanied by several of these skeletal complications including spondylodiscitis.

Case report

A 29-year-old man with GD was seen for his complaints of low back pain, fever, and hypotension. He described the back pain as insidious but progressively increasing and radiating to both legs while he was standing. He also complained of fatigue, lack of appetite, and weight loss for the last 4 months. He had been initially admitted to another center with the same complaints 3 months ago. At that time, his body temperature had been 39°C, and the blood biochemistry profile had been normal; however, the erythrocyte sedimentation rate (ESR) (110 mm/h) and C-reactive protein (CRP) (36.7 mg/l; normal 0–0.5 mg/l) level was increased. Accordingly, he had been given several empirical antimicrobial treatments for a likely diagnosis of spondylodiscitis. His current physical examination revealed a body temperature of 38.6°C, tenderness, and paravertebral muscle spasm in the thoracolumbar region. The laboratory tests were as follows: ESR 115 mm/h; CRP 39.2 mg/l (normal 0–0.8 mg/l), leukocytes 14 600/mm, hemoglobin 8.6 g/dl, platelets 161 000/mm. Liver enzyme levels were elevated, but kidney function tests were normal. Biochemical bone markers were as follows: Ca 7.83 mg/dl (normal 8.6–10.2 mg/dl), phosphorus 3.2 mg/dl (normal 2.3–4.7 mg/dl), ionized calcium 1.01 mmol/l (normal 1.15–1.29 mmol/l), parathyroid hormone 28.7 pg/ml (normal 9.5–75 pg/ml), alkaline phosphatase 283.4 U/l (normal 35–129 U/l), bone-specific alkaline phosphatase 88.2 µg/l (normal 11.6–20.1 µg/l), 24-h urinary Ca excretion 414.3 mg/day (normal 100–300 mg/day), urinary deoxypyridinoline 141 nM DPD/mMcre (normal 2.8–5.6 nM DPD/mMcre). Bone mineral densitometry yielded an L1–L4 T-score of −2.73 and a femoral neck T-score of +0.13. No oligoclonal band was observed during serum and urine protein electrophoresis. Mepheridin and diclofenac were started for his severe back pain, and a rigid thoracolombar brace was recommended during ambulation.

Because of his persistent back pain and the fever, radiological evaluations for the thoracolumbar vertebrae were performed and were consistent with involvement of the spine due to GD and concomitant spondylodiscitis (Fig. 2). Although we could not determine any infectious pathogen (including tuberculosis or brucella) from blood cultures, intravenous clavulanic acid and ampicillin were empirically started based on the clinical and radiological findings. Bone marrow biopsy displayed diffuse histiocytic infiltration concordant with a storage disease. Owing to the patient’s aggravated pain, gabapentin 3 × 600 mg was given and
A morphine pump had been put in place. Computed tomography (CT) of the thoracolumbar spine was repeated (Fig. 2B) along with a radiological survey (Fig. 3). Due to ongoing infection despite aggressive antibiotic treatment, glucocerebrosidase enzyme replacement therapy (ERT) was prescribed, and surgical intervention was postponed.

After the infection subsided and his medical condition stabilized, the patient was taken to surgery. T12L1 corpectomy and cage fixation were applied via an anterior approach. Posterior instrumentation and fusion were performed using pedicle screws (bilaterally at T69 and L13 vertebrae) and two rods. Autogenous and allogeneic grafts were implanted both using anterior and posterior approaches. On postoperative day (POD) 1, the patient could sit with a rigid thoracolumbar brace; and on the next day, he was mobilized. Currently, the patient is under ERT without any symptoms or signs of infection.

**Discussion**

Gaucher's disease is an autosomal recessive lysosomal storage disorder seen in association with more than 300 mutations of the β-glucocerebrosidase gene, located on chromosome 1q21. It is characterized by a wide spectrum of manifestations that generally take place owing to initial hematological abnormalities secondary to hypersplenism. Other gastrointestinal complications, including hepatomegaly, cirrhosis, ascites, esophageal varices, hepatic carcinoma, and cholelithiasis, may also ensue. Depending on the status of the patient, enzyme replacement and substrate reduction remain the mainstay of treatment.

Musculoskeletal manifestations are probably the most disabling aspects of the disease, leading to impaired movement and pain.