Gaucher disease presenting with vertebral compression fractures and vertebral osteonecrosis

G. Clinckemaillie¹, A. Larbi², P. Omoumi², J. Manelle¹, B. Dallaudière²,⁴

A 42-year-old man with no relevant medical history complaining of spontaneous thoracolumbar back pain was referred for an MRI of the spine after initial radiographic evaluation had demonstrated several compression fractures.

In addition to confirming the dorsal and lumbar compression fractures and some degenerative changes, the MRI demonstrated multifocal bone marrow anomalies with well-defined geographical zones of low signal on T1-weighted images (Fig. A) and intermediate to high signal on STIR images (Fig. B) in dorsal and lumbar vertebral bodies as well as in the sacrum (closed arrows in both figures). In some vertebral bodies, and in particular in the sacrum, a subtle double-line sign was present (arrow head in Figure B), suggesting the diagnosis of osteonecrosis.

After ruling out common causes of multiple vertebral osteonecrosis and taking the Ashkenazic Jewish descent of the patient into account, the diagnosis of Gaucher disease was proposed. Genetic testing confirmed the diagnosis. Supportive therapy was quickly started after the diagnosis was confirmed in this patient. Enzyme replacement therapy (ERT) might be offered in nonneuropathic GD but its high cost and the variable response make it necessary to define appropriate clinical indications. ERT was not started in this patient.

Comment

Gaucher disease (GD) is an autosomal recessive metabolic disorder caused by deficiency of the enzyme glucocerebrosidase and hence characterized by the abnormal deposition and accumulation of glucocerebroside in the lysosomes of reticuloendothelial cells (“Gaucher cells”). These cells can accumulate in the spleen, liver, kidneys, lungs, brain and bone marrow. Symptomatology, organ involvement and clinical course vary greatly among affected patients. Despite this heterogeneity, three basic clinical forms have been identified, based on the degree of neurological involvement. Most patients have the nonneuropathic form, referred to as type 1 GD or adult GD; types 2 and 3, respectively acute and subacute neuropathic forms, occur in the remainder of the patients. Systemic symptoms are more common than neurological involvement in patients with GD.

Accumulated Gaucher cells in the bone marrow replace the normal cellular population, causing ischaemia and as a result edema, which is clinically accompanied by pain. Bone adjacent to marrow infiltration may exhibit cortical thinning or scalloping, osteopenia and deformity. Further accumulation of Gaucher cells leads to fractures, necrosis, and less frequently osteomyelitis. In the axial skeleton, multiple compression fractures can be observed, leading to kyphosis and eventually to spinal cord compression. The involved vertebral bodies may become completely flattened. Less frequently, H-shaped, step-like defects in the vertebral bodies may be observed.

On MRI, the described infiltration of the bone marrow with accumulating Gaucher cells will be manifested as a decrease in T1- and T2-signal intensity, and may display a homogeneous or heterogeneous pattern. MRI is the most sensitive imaging modality to demonstrate osteonecrosis. The findings will depend on the stage of necrosis, as described in the Mitchell classification, based on the signal characteristics within the central lesion. Class A is early osteonecrosis, with signal intensity characteristics analogous to those of fat, being high signal intensity on T1-weighted images and intermediate signal intensity on T2-weighted images. Class B is manifested as a high signal on T1- and T2-weighted images, comparable to signal intensity characteristics of blood. Class C demonstrates signal intensity characteristics similar to those of water, with low signal on T1- and high signal on T2-weighted images. Class D is the most advanced stage and demonstrates low signal intensity on both T1- and T2-weighted images, resulting from fibrous tissue proliferation. In GD, areas of necrosis are typically multiple, with well-defined, serpiginous margins, as was the case in the presented patient.

Reference