Type I Gaucher disease: extraosseous extension of skeletal disease

Abstract  Objective. To investigate the frequency and morphology of extraosseous extension in patients with Gaucher disease type I.

Design and patients. MRI examinations of the lower extremities were analyzed in 70 patients with Gaucher disease type I. Additionally, the thoracic spine and the midface were investigated on MRI in two patients.

Results. Four cases are presented in which patients with Gaucher disease type I and severe skeletal involvement developed destruction or protrusion of the cortex with extraosseous extension into soft tissues. In one patient, Gaucher cell deposits destroyed the cortex of the mandible and extended into the masseter muscle. In the second patient, multiple paravertebral masses with localized destruction of the cortex were apparent on MRI in two patients.

Conclusions. Extraosseous extension is a rare manifestation of Gaucher bone disease. While an increased risk of cancer, especially hematopoietic in origin, is known in patients with Gaucher disease, these extraosseous benign manifestations that may mimic malignant processes should be considered in the differential diagnosis of extraosseous extension into soft tissues. A narrow neck of tissue was apparent in all cases connecting bone and extraosseous extensions.

Key words  Gaucher disease · Bone disease · Extraosseous Gaucher disease · Bone marrow imaging · MRI

Introduction

Gaucher disease is an inborn error of glycosphingolipid metabolism and is the most frequent lysosomal storage disorder [1]. It is caused by an insufficient amount of glucocerebrosidase (glucosylsphingosidase) activity with secondary accumulation of glucocerebrosides within the lysosomes of macrophages [2, 3]. The storage disorder produces a multisystem disease characterized by progressive visceral enlargement and gradual replacement of bone marrow with lipid-laden macrophages. Symptomatic anemia, coagulation abnormalities, hepatosplenomegaly and structural skeletal changes occur at some point during the course of illness in most patients.

Skeletal involvement is a major cause of morbidity, afflicting up to 75% of patients, but varies widely between and within individual patients. Bone lesions range from mild osteopenia, medullary expansion and remodeling defects (e.g., Erlenmeyer flask deformity) to osteonecrosis of the femoral or humeral heads and spinal cord compression from vertebral collapse [4, 5]. Extraosseous manifestation of Gaucher disease is supposed to be extremely rare [6, 7]. We report four patients with Gaucher disease type I demonstrating extraosseous extension or medullary expansion with protrusion of the cortex.

Since 1991, patients with Gaucher disease type I have been examined clinically and radiologically by an interdisciplinary study group. MRI of the lower extremities in
Fig. 1 Case 1. T1-weighted axial spin-echo image of the midface after gadolinium enhancement (TR/TE: 625/25 ms; slice thickness 5 mm) shows a low signal intensity mass in the right masseter muscle with a contrast-enhancing capsule (*black arrows*). *M* right masseter muscle

Fig. 2 Case 1. On T2-weighted axial turbo-spin-echo (TSE) image of the right midface (TR/TE: 2200/100 ms; slice thickness 3 mm) Gaucher cell extension appears hyperintense, indicating fluid or proteinaceous content within the mass. Note the narrow neck of tissue between the lateral cortical mandible and the tumor (*white arrows*). *P* right parotid gland

Fig. 3 Case 1. High-power photomicrograph of the extraosseous soft tissue mass with accumulation of glucocerebroside-laden histiocytes and plasma cell infiltrates. (PAS, original magnification ×870)

Fig. 4 Case 2. Contrast-enhanced CT scan of the chest demonstrates paravertebral masses with homogeneous, strong contrast enhancement (*white arrows*)

Fig. 5 Case 2. Post-myelography CT scan shows an osteolytic lesion within a thoracic vertebral body (*black arrowhead*) and a lytic intracortical lesion (*small black arrows*)