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CASE REPORT

Cauda equina syndrome in a patient diagnosed with type 1 Gaucher disease: a rare case

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Abstract

Background Gaucher disease is a rare hereditary glycolipid storage disease. One of the rare complications is neurodeficits due to vertebral involvement.

Case presentation An 18-year-old female patient presented to the outpatient clinic with cauda equina syndrome due to sacral involvement of type 1 GD. Bilateral laminectomy via posterior approach without posterior stabilization was performed.

Conclusion Maximum excision of the mass avoiding destabilization of the spinal column can provide long-term vertebral stability and improvement in neurodeficits.

Keywords Gaucher · Cauda equina · Sacral mass

Introduction

Gaucher disease (GD) is a rare, autosomal recessive hereditary glycolipid storage disease caused by a mutation in the β-glucocerebrosidase 1q21 gene and the resulting deficiency of the enzyme glucocerebrosidase [1, 2]. Type 1 is the most common form. The most frequent clinical findings of type 1 GD are related to hepatosplenomegaly, thrombocytopenia, anemia, and long bone lesions [3, 4]. The vertebrae are rarely affected [4]. In cases with vertebral involvement, the disease can also lead to clinical findings associated with spinal cord and nerve root involvement [5, 6]. Here, we present a case of cauda equina syndrome due to sacral involvement in a patient with type 1 GD.

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Case

An 18-year-old female patient presented to the outpatient clinic with complaints of progressive leg numbness, weakness, and urinary incontinence starting 45 days earlier. Also, she had a complaint of low back pain which was partially reduced by the analgesics she felt at the time of the last 6 months. The patient had no history of trauma, cancer, or spinal pathology, but had been examined 4 years earlier for recurrent bone fractures, thrombocytopenia, splenomegaly, and hepatomegaly and diagnosed with type 1 GD at another health center. After the first diagnosis, the patient had gotten cerezyme 60 U/kg ERT (enzyme replacement therapy) once every 2 weeks. She has been continuing to get 15 U/kg once every 2 weeks since 2 years. Neurological examination revealed that the patient had no neurodeficit in the upper extremities, was paraparetic (1/5 motor strength) and hypoesthetic in the lower extremities, and exhibited hypoactive deep tendon reflexes and lack of sensation of the saddle area. Urgent spinal magnetic resonance imaging (MRI) was performed. The lesions had irregular borders and caused extradural compression in the lumbosacral region, resulting in bone destruction (Fig. 1). Computed tomography (CT) imaging showed GD-related bone marrow involvement in all vertebrae (Fig. 2). Based on these findings, the patient without anemia and thrombocytopenia in laboratory tests was operated on the first day of admission. Assisted by neuromonitoring, bilateral laminectomy via posterior approach was performed, followed by subtotal excision of the

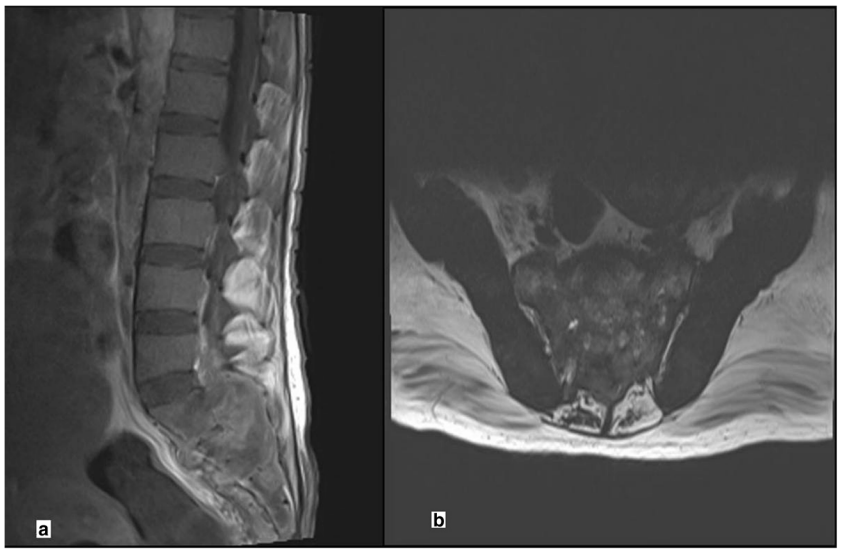

Fig. 1 **a** View of the mass in the sagittal sectioned MR image with contrast before surgery. **b** View of the mass in the axial sectioned MR image with contrast before

extradural mass for spinal cord decompression. Posterior stabilization was not done. She was discharged on the fifth day after the operation. Hematology and endocrinology did not suggest any treatment except the same dose ERT after discharge. At postoperative 6 months, the patient's lower extremity paraparesis had improved to 3/5 motor strength, and anal tone and sensitivity had returned to normal. No recurrence was observed during 18 months of follow-up (Fig. 3).

Discussion

There is no direct central nervous system involvement in type 1 GD. Neurological symptoms are mainly due to spinal cord and nerve root compression caused by vertebral collapse. Vertebral fractures are believed to occur as a result of bone fragility caused by the accumulation of fat cells (Gaucher cells) in the bone marrow [7]. More rarely, spinal canal compression and consequent neurodeficit can develop in the absence of bony compression, due to mass effect exerted by Gaucher cells accumulating in the epidural space after extravasation from the bone marrow, as was observed in case reports in the literature [5, 8]. However, unlike the case reported, in our patient, the epidural mass was located in the sacral region with epidural space and caused cauda equina syndrome. It should be kept in mind that hematomyelia associated with thrombocytopenia caused by type 1 GD may also lead to neurodeficit [7].

According to our review of the literature, vertebral involvement occurs primarily in the thoracolumbar region, while involvement of the lumbosacral region is less common [9].

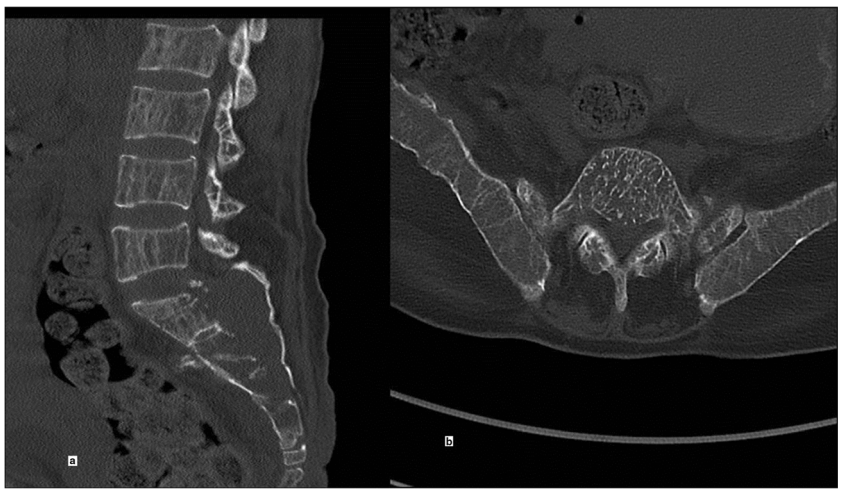

Fig. 2 **a** View of the mass in the sagittal sectioned CT image before surgery. **b** View of the mass in the axial sectioned CT image before surgery

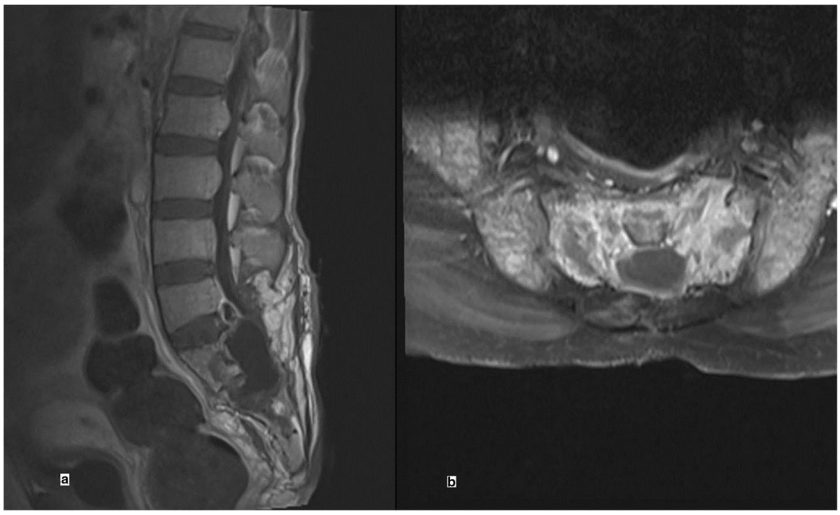

Fig. 3 **a** View of the mass place in the sagittal sectioned MR image with contrast after the 18th month of surgery. **b** View of the mass place in the axial sectioned MR image with contrast after the 18th month of surgery

The thoracic vertebrae are generally affected in adolescent cases [5, 10]. There was also reported that 16 of 105 adult type 1 GH patients had vertebral involvement, and that the thoracolumbar region was affected in all of those cases [11].This involvement is shown to lead to kyphotic deformity due to vertebral compression in the thoracolumbar region especially and to bring about destruction of the sacral vertebra in the sacral region (Table 1).These vertebral deformities can be easily visualized using X-ray and CT imaging. It usually shows heterogeneous contrast enhancement on MRI. It should be remembered that lesions may also be cystic.

Treatment is conservative for patients with severe pain due to vertebral involvement. There are reports that skeletal healing may be possible, especially with long-term and high-dose ERT. Also, it is stated that the ERT is generally an effective response to visceral tissues [12].However, for patients with vertebral collapse, the degree of kyphosis and spinal cord compression caused by vertebral collapse are the factors in deciding to perform surgery. The pressure being exerted on the spinal cord by the Gaucher cell mass and the neurodeficit exhibited by the patient also lead to surgical intervention. The goal of surgery is to remove as much of the lesion causing the neurodeficit as possible before the neurodeficit progressed, while avoiding sacral vertebral instability. We performed neuromonitoring-guided maximal mass resection while sparing the sacral cortex to achieve this. Because of the clinical symptoms being caused by the epidural compression, we opted to first eliminate the pressure. Examination of the patient's preoperative imaging showed that none of the vertebrae in that region were healthy (Fig. 2).Our approach was validated by the reduction of neurodeficit and the lack of instability during the 18-month follow-up period (Fig. 3).

Conclusion

Although sacral involvement is rare in Gaucher disease, it should not be overlooked in the differential diagnosis of sacral masses. Patients with neurodeficits resulting from spinal compression associated with Gaucher cell-induced lesions should be treated with maximum safe resection, but posterior stabilization may not be necessary.

Table 1 Published examples of Gaucher's disease related to vertebral involvement

	Lesion area	Spinal pathology	Surgery method
Hamlat et al. (1 patient)	Sacral	Intradural cyst	Laminectomy
Hermann et al. (3 patients)	Thoracic (T11)	Kyphotic deformity	Stabilization
	Thoracic (T12)	Collapse	Conservative
	Thoracic (T2–T3)	Epidural lesion	Stabilization
Javier et al. (16 patients)	Thoracolumbar (T3–L5)	Vertebra fracture	Stabilization
Kocher et al. (1 patient)	Thoracic (T8)	Kyphotic deformity	Stabilization

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