Gorham-Stout Disease in a Middle-Aged Patient Treated by Posterior Lateral Fusion: A Case Report and Literature Review

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Gorham-Stout disease (GSD) is a rare progressive osteolytic disease with intraosseous lymphangiomatosis. A previously healthy 55-year-old woman presented with left leg hypoesthesia and upper back pain. k pain, back pain, and bilateral shoulder pain. In radiologic examination, there was an osteolytic lesion in T3, 4, 5, 6 vertebra. And a dislocation in the T3/4 vertebrae and a compression fracture in the T4 vertebra were founded. She was treated with posterolateral fusion (PLF) from C7 to T7. For 1 year, she had no specific symptoms but a new osteolytic lesion was found on thoracic T8 on 1 year follow-up magnetic resonance imaging. Radiotherapy was performed with 40 Gy, and chemotherapy was performed with denosumab. This report described a case of GSD that involved the thoracic spine and was appropriately managed. In the present case of a middle-aged Asian woman, correction of severe thoracic instability and prevention of neurologic compromise were accomplished through PLF from C7 to T7. Recurrence was observed 1 year later, but it was appropriately treated with neoadjuvant therapy.

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INTRODUCTION

Gorham-Stout disease (GSD), also known as "vanishing bone disease" or "massive osteolysis", is a rare disease of unknown etiology that is characterized by progressive osteolysis and absorption with mono- or poly-osteolytic lesions⁷. Gorham disease was first described in 1838; since then, more than 300 cases have been reported, with approximately 60 cases related to the spine. One common complication of the disease is the development of chylothorax, which occurs in approximately 20% of patients and results in a high mortality rate¹³. No standard diagnostic tool or therapy exists for the disease so that it can be easily misdiagnosed. Depending on disease severity and extension of organ involvement, commonly used treatments include surgery, radiotherapy, and pharmaceuticals. GSD can be diagnosed at any age, but it is most commonly observed in children and young adults (the average age of diagnosis is 25 years). This study report management of a middle-aged patient with GSD on thoracic spine combined thoracic vertebral dislocation.

CASE REPORT

A previously healthy 55-year-old Asian woman presented with left leg hypoesthesia and upper back pain. Before being admitted to the hospital, she visited a local clinic and was diagnosed with fibromyalgia with neck pain, back pain, and bilateral shoulder pain. She had a history of a fall 2 weeks prior to admission, after which

symptoms were aggravated. One day before visiting our institution, she noticed a change in her gait and hypoesthesia of her left leg. She had no family history of hereditary disease. On physical examination, she exhibited tenderness in the upper back and a slight loss of left leg sensation (about 60% remained). Thoracic spine computed tomography (CT) revealed a dislocation in the T3/4 vertebrae and a compression fracture in the T4 vertebra. Magnetic resonance imaging (MRI) scans showed a combined pathological fracture of T4 with anterior subluxation of T3 over T4, causing compressive myelopathy/cord contusion. In addition, the T1 to T6 posterior elements and heads of the left 3rd to 5th ribs showed a hypoplastic appearance with or without osteolysis. A whole-body bone scan demonstrated focal uptake at the T4 area due to the known compression fracture and mild uptake at the spinous process area of T6 due to another fracture (Fig. 1). Blood analyses, including multiple myelopathy tests, indicated normal biochemical levels. She was admitted through the emergency room with T3/4 dislocation and cord compression.

In the present case, cord compression and vertebral dislocation required surgical treatment, and the plan was posterolateral fusion (PLF) from C7 to T7. With the patient under general anesthesia, the surgery was performed in a patient in the prone position of Jackson table. After confirming the surgical site on the radiograph, a dorsal midline incision was made. The subcutaneous fat layer and fascia were dissected and the muscles were separated from the vertebral arch plate. T3 and T4 total laminectomy were performed for decompression of the spinal cord. The bones were very loose

Fig. 1. (A) Preoperative thoracic spine computed tomography imaging shows a dislocation at T3 and T4 level. (B) Preoperative thoracic spine magnetic resonance imaging T1 contrast enhance sagittal image shows a combined pathological fracture of T4 with anterior subluxation of T3 over T4, causing compressive cord contusion. (C) Preoperative whole-body bone scan shows focal uptake appeared at T4 area. (D) Preoperative positron emission tomography shows irregular osteolytic and hypoplastic change at T3, 4, 5, and 6 area.

Fig. 2. (A, B) Postoperative lateral and anteroposterior radiograph of the thoracic spine at the follow-up of 4 months after surgery, showing a posterolateral fusion on C7-T7. (C) Computed tomography of the thoracic spine at the follow-up of 4 months after surgery, showing a stabilization of C3/4 vertebral body. (D) Magnetic resonance imaging of the thoracic spine showing a cord decompression at the follow-up of 4 months after surgery.

and, unlike normal bones, they were very thin. Because the bone was very weak and reduction was needed for correcting T3/4 dislocation, pedicle screw insertion was performed up to C7-T7 and bone fusion was conducted with allogeneic materials. A hemovac drain was placed on the surgical site and the wound was closed along the edge. The surgery was completed uneventfully.

After the operation, her symptoms improved. A Minerva jacket was prescribed to prevent kyphotic deformity and to support the PLF. Postoperative pathological examination of the bone tissue (T3

and 4 vertebral bodies) biopsy revealed GSD with thin-walled vascular proliferation replacing bone marrow and laminated trabecular bone with fibrous stroma. Two weeks after surgery, chest CT showed bilateral pleural effusions, and the chest was drained on both sides. The drain catheter inserted for the pleural effusions showed that the effusions were exudate, not chylothorax. After 2 weeks, both pleural effusions improved and the drain was removed. Since there is no gold standard for GSD, in this patient, the active phase passed by over 50 years of age, and since there was no other special disease

Fig. 3. (A, B) Postoperative lateral and anteroposterior radiograph of the thoracic spine at the follow-up of 1 year after surgery, showing a posterolateral fusion on C7-T7. (C, D) Magnetic resonance imaging of the thoracic spine showing a new osteolytic lesion on T8 left vertebral body, left pedicle on both T2 weighted image and T1 enhance image at the follow-up of 1 year after surgery.

before, chemotherapy and radiotherapy were not performed after surgery, and it was decided to follow up. The 4 months after surgery, thoracic spine X-ray and CT scan showed successful reduction and stabilization of the thoracic spine deformity (Fig. 2). A new osteolytic lesion was found involving T8 left vertebral body, left pedicle, bilateral transverse process, left 8th and 9th rib head on 1 year follow up MRI (Fig. 3). Radiotherapy was performed with 40 Gy with denosumab. There is no recurrence during 8 months after the radiotherapy (Fig. 4).

DISCUSSION

GSD is also known as vanishing bone disease, massive osteolysis, and disappearing bone disease. Gorham et al.³ reported 24 cases and defined the disease as a syndrome in 1954. Although the exact pathophysiology of GSD is not known, the correlation between lymphatic proliferation and gradual osteolysis is well-established. Gorham and Stout² originally posited angiomatosis caused by hyperemia, interrupted bone metabolism, and altered balance to favor osteoclastic activity. GSD presents no hereditary or sex tendency and can occur at any age, although it is more common in adolescents and young adults⁶. As in our case, it is rarely found in people over 50. GSD can involve any part of any bone; however, the skull, shoulder, cervical vertebrae and pelvis are the most frequently involved sites¹⁰. Especially, spinal involvement can lead to instability and neurologic impairment. In our case, GSD occurred in the thoracic spine, and neurologic abnormality occurred due to dislocation. Treatment of spinal GSD requires close observation and multi-disciplinary clinical decision-making. Because no standard therapy exists, care of patients with GSD is case specific. Some available

Fig. 4. T2 magnetic resonance imaging thoracic spine at the follow-up after 8 months of end of radiosurgery.

treatment modalities include pharmaceutical therapy, chemotherapy, radiotherapy, and surgical intervention⁴. Between 1947 and 2016, 59 cases of spinal GSD were reported in the literature. Of these 59 cases, 49% involved the cervical spine, 46% involved the thoracic spine, 17% involved the lumbar spine, and 5% involved the sacral spine¹². In almost all cases, multiple spine levels were involved. Among the 59 patients, 28 underwent surgical intervention¹². Spinal lesions can be managed by radiation therapy, vertebral osteotomy,

braces, balloon vertebroplasty with cement augmentation, or spinal fusion with bone graft to correct instability and prevent neurologic impairment^{1,8,9)}. Tateda et al.¹²⁾ reported that chemotherapy before surgical correction was effective. GSD patients can gain better surgical outcomes through neoadjuvant chemotherapy targeting involved tissue. In our case, neoadjuvant therapy was not administered after surgery. This is because, based on the age of the patient, it was judged that the patient's GSD state had passed the active phase. A new lesion was found 1 year later, and radiotherapy were performed, but there is a regret that it would have been better if performed immediately after surgery. Chylothorax can occur when this disease involves the pleura or thoracic duct, and several studies report an association between GSD and chylothorax^{5,11)}. Bilateral chylothorax is usually lethal and can cause gradual respiratory failure with an increased mortality rate. Our case showed bilateral pleural effusions 2 weeks after surgery, but the effluent was proven to be exudate, not chylothorax. Further studies are required to evaluate what causes an exudative condition.

CONCLUSION

This report described a case of GSD that involved the thoracic spine and was appropriately managed. In the present case of a middle-aged Asian woman, correction of severe thoracic instability and prevention of neurologic compromise were accomplished through PLF from C7 to T7. Recurrence was observed 1 year later, but it was appropriately treated with chemotherapy and radiotherapy.

CONFLICTS OF INTEREST

No potential conflict of interest relevant to this article was reported.

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