Utility of the spinal instability neoplastic score to identify patients with Gorham-Stout disease requiring spine surgery

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ABSTRACT

Background: Gorham-Stout disease (GSD) is a rare syndrome presenting with progressive osteolysis which in the spine can lead to cord injury, instability, and deformity. Here, the early spine surgery may prevent catastrophic outcomes.

Case Description: A 25-year-old male with GSD involving the T2 to T6 levels presented with acute traumatic kyphoscoliosis at T3 and T4 and left lower extremity paraparesis. The CT scan 4 weeks before this showed progressing osteolysis versus the CT 5 years ago. Unfortunately, the patient underwent delayed treatment resulting in permanent neurological sequelae. Surgery included a laminectomy and vertebrectomy of T3/T4 with instrumented fusion from T1-10. The use of the spinal instability neoplastic score (SINS) is a useful tool to prompt early referral to spine surgeons.

Conclusion: We recommend using the SINS score in GSD patients who develop spinal lesions to prompt early referral for consideration of surgery.

Keywords: Gorham-Stout, Instability, Reconstruction, Spine

BACKGROUND AND IMPORTANCE

Gorham-Stout disease (GSD) is rare; only approximately 300 cases have been reported.[11] The condition is attributed to the nonneoplastic proliferation and dilation of lymphatic channels resulting in destruction of the cortical ribbon in an average of 7.5 bones by the time of clinical presentation/diagnosis; these include (in descending frequency) ribs, cranium, clavicle, cervical spine, thoracic spine, humerus, and femur.[12,10,13] Although the osteolysis is progressive, it is occasionally spontaneously arrested or diminished following medical and/or radiation treatment.[11,12,14]

Spinal involvement has been reported in 75 patients, 33 of which required surgery for fracture or deformity.[12,15] Here, we report a patient newly diagnosed with GSD who acutely presented with a T3/4 spinal deformity that was not managed in a timely fashion (i.e., C3, T4 corpectomy/posterior fusion T1-T10) and resulted in a permanent paraparesis.
CLINICAL PRESENTATION

A 25-year-old male with GSD presented with traumatic kyphoscoliosis centered at the T3-T4 level.[11] Previously, serial MR studies showed asymptomatic progressive fatty marrow replacement of the T4-T6 spinous processes and T2-T6 vertebral bodies/posterior elements; nothing had changed over 3 years [Figure 1a-c]. Four weeks before admission, the patient presented with a T3 spinous process fracture, and the CT showed osteolysis of the T2-T6 vertebral bodies and left T3-T8 facet joints; these findings had progressed compared to a CT of the thorax 5 years prior. On the current admission, the patient now had a significant paraparesis with a T4 sensory level. The MR showed spinal canal stenosis at T3-T4 with hyperintense signal in the cord on T2 sequences [Figure 1d]. The CT demonstrated a fracture of the T3 inferior endplate and T4 superior endplate with extension through the right superior facet and left inferior facet resulting in destruction of the posterior elements and 53° of kyphosis with levsoscoliosis [Figure 2a and b].

The patient underwent a partial vertebrectomy at T3 and T4 and posterior instrumented fusion from T1 to T10 [Figure 2c]. The right T4 nerve root was sacrificed during the vertebrectomy, and a traumatic dural laceration sustained as a result of the fracture was repaired with an autologous duraplasty. The patient made an uneventful recovery, except for a persisting left pleural effusion, a known complication of GSD that was present preoperatively and unrelated to surgical intervention. At 3-month follow-up, the entire left leg was numb more consistent with a neurological level of T12 and below. A zone of partial preservation was observed from T6 to T12.

DISCUSSION

The timing of surgery in the treatment of GSD is challenging. Operating while the disease is active is not a definitive management because instability or neurological deficits may occur as osteolysis progresses.[8,15] As a result, the trend has been to conservatively treat spinal lesions and carry out fusion only after osteolysis has ceased.[6,15] The progressive nature of the disease necessitates multiple revision surgeries in half of reported cases and complications such as neurologic deficit, chylothorax, and a high rate of mortality are common.[1,4,8,9,15] In patients with spinal lesions, it is critical for the treating physician to identify patients at risk of catastrophic instability. These patients undoubtedly benefit from the early involvement of a spine surgeon to assess stability and instigate prompt access to surgical care.

The spinal instability neoplastic score (SINS) was initially developed to determine instability of the vertebral column due to metastatic tumor burden visible on CT. It is easy to use for nonsurgical personnel in an inpatient or outpatient setting using standard investigations to identify and quantify spinal instability with patients scoring “potential instability” or “instability” being referred for an urgent surgical opinion. If employed in cases of GSD, the score could serve as a tool to trigger early consultation with a spine surgeon before symptomatic spinal instability or neurological compromise.[7] Before the pathologic fracture, this patient had a SINS of 7 (location: 1; pain: 3; bone lesion: 2; radiographic spine alignment: 0; vertebral body collapse: 0; and posterior spinal element involvement: 1), suggesting a potentially unstable spine. If identified at the time of the T3 spinous process fracture, consultation with spine surgery may have prevented his subsequent injury and complex procedure.

Figure 1: Annual MR imaging of the thoracic spine over 3 years (a-c) demonstrates stability of the increased signal intensity of the T2-T6 vertebral bodies, which is consistent with the fatty marrow replacement seen in Gorham-Stout disease. Postinjury imaging (d) demonstrates severe kyphosis at T3-T4. Acute cord signal change is evident, in keeping with acute myelopathy (d).
CONCLUSION

In GSD patients with known spinal lesions, we recommend serial evaluation of stability using the SINS and involving a spine surgeon early to prevent progressive instability, deformities, and neurologic deficits.

Declarations of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Conflicts of interest

There are no conflicts of interest.

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