Gorham's Disease in a Patient with Traumatic Spinal Cord Injury: Case Report and Review of the Literature

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Gorham's disease is a rare entity that has been sparsely covered in the medical literature, and its pathophysiology remains poorly understood. We present the case of a 22-year-old man who sustained a traumatic T6 American Spinal Injury Association Impairment Scale B paraplegic injury complicated by a complaint of shoulder pain during his acute rehabilitation stay. He was found to have osteolysis of the distal right clavicle (Gorham's disease). He was treated conservatively with nonsteroidal anti-inflammatory drugs and relative rest and experienced good functional outcome. Although the differential diagnosis for shoulder pain in the paraplegic patient during acute rehabilitation is extensive, it is important to consider less common but still important etiologies such as Gorham's disease. **Key words:** bone health, Gorham's disease, shoulder pain, spinal cord injury

Gorham's disease is a rarely encountered clinical phenomenon that has been scarcely reported in the literature. It is known by several other names including disappearing bone disease, essential osteolysis, massive osteolysis, idiopathic massive osteolysis, progressive massive osteolysis, Gorham's syndrome, massive Gorham osteolysis, Morbus Gorham-Stout disease, vanishing bone disease, and weightlifter's shoulder. There is an adage that the more names a disease has, the less is known about it. Gorham's disease is no exception. Despite its first description in 1954 by Gorham et al,¹ it remains a poorly understood phenomenon.² The disease does not appear to have neurological, infectious, or oncological origins,² but rather appears to be related to lymphangiomatosis³ in which bone is replaced by fibrovascular connective tissue. There is a predilection for intramembranously formed bone.⁴ As of 2013, only 200 cases had been reported in the literature, and adequate epidemiological statistics are unreliable. The vast majority of the cases reported involve only a single bone.^{4,5} The risk factors for the disease are unknown, but it appears that it is more common in persons under the age of

40 years. Some investigators believe that the disease is more common in men; others state that the ratio is closer to 1:1.^{2,3,6} There does not appear to be a predilection to disease onset after major traumas, such as spinal cord injury (SCI).

Case Report

We report the case of a 22-year-old man with a previously unremarkable medical history who sustained a T8 vertebral fracture in a motor vehicle collision, resulting in T6 American Spinal Injury Association Impairment Scale (AIS) B paraplegia on the Abbreviated Injury Scale. Additional injuries included bilateral pulmonary contusions and hemothorax requiring a right video-assisted thoracoscopic surgical (VATS) procedure with drainage and chest tube placement by cardiothoracic surgery. After the patient was stabilized in acute care and after the chest tubes had been removed, he was transferred to acute rehabilitation. During his acute rehabilitation stay, he was maintained in a thoracolumbosacral orthosis (TLSO). After 2 weeks in rehabilitation, he began to complain of an insidious onset of moderate, dull, and

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Top Spinal Cord Inj Rehabil 2016;22(1):79–82
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www.thomasland.com

doi: 10.1310/sci2201-79

localized shoulder pain that worsened with activity. A plain film was obtained and demonstrated marked atrophy of his right distal clavicle (see **Figure 1**), suggestive of Gorham's disease. He was treated conservatively with nonsteroidal

anti-inflammatory drugs (NSAIDs) and rest, and there was no extension of his disease. In follow-up, it was found that he manages well without any pharmaceutical or surgical interventions and has regained functional use of his shoulder.

Figure 1. Anteroposterior view of the right shoulder during internal rotation. Note the distal clavicular lucency (arrow) indicative of osteolysis with preservation of the acromion.

Discussion

Shoulder pain is a commonly experienced phenomenon for the paraplegic patient and has many potential etiologies. Musculoskeletal injuries related to initial trauma or therapy remain the most common, but development of syring, neuropathic pain resulting from nerve injury (eg, brachial plexopathy, cervical radiculopathies, axillary and suprascapular neuropathies), and referred pain from organ injury (such as gallbladder disease or myocardial infarction) are also possible.⁷ A complete review of shoulder pain in paraplegic patients is beyond the scope of this article, and the reader is referred elsewhere.⁸⁻¹⁰ We simply stress the importance of formulating a wide differential diagnosis. Since shoulder pain is such a common phenomenon in patients with SCI (both in tetraplegia and paraplegia), facility with the wide differential of shoulder pain is paramount for a health care provider who treats patients with SCI. Less common etiologies should be considered and ruled out when possible. Gorham's disease is a potential cause of shoulder pain, and an accurate and early diagnosis may save the patient from extensive workup and invasive testing and treatment, while providing reassurance of a favorable prognosis. Because patients with SCI rely heavily on their upper extremities for functional mobility and performance of activities of daily living, early recognition and treatment of upper limb pain syndromes takes on additional importance.

Gorham's disease has been reported in most bones of the body and can occur in both the appendicular and axial skeleton. According to Ahlmann et al, the most commonly affected bones include the "scapula, clavicle, humerus, pelvis, femur, ribs, skull, maxillofacial bones, mandible, vertebral bodies, hand, and foot."^{4(p1)} Histologically, Gorham's disease is characterized by the proliferation of hemangiomatous or lymphangiomatous cells without cellular atypia, leading many to believe that this entity is related to lymphangiomatosis.³ On the cellular level, osteoclast precursor cells may become more sensitive to humoral factors, which stimulate maturation and development of mature osteoclasts and subsequent bony resorption.¹¹

Clinical symptoms are variable but are generally referable to the site of disease. Thoracic disease (in the ribs, sternum, spine) can lead to a potentially devastating chylothorax secondary to direct lymphangiomatic extension into the thoracic duct. The disease, however, can present acutely or insidiously, with or without local erythema and/or edema, and the description of pain can vary. The symptoms may or may not be associated with progressive weakness or limitation in active or passive range of motion. Serious complications are rare. Death can occur as a result of respiratory failure when thoracic or pleural tissues are involved and with skull involvement. SCI has been reported as a result of vertebral involvement. Osteomyelitis is extremely rare but has also been reported.²

Diagnosis is made by obtaining a history excluding other potential etiologies, by performing a physical exam evaluating for patterns of tenderness and restrictions in range of motion, and by radiographic examination. Because of the osteolytic components, the bone appears to "disappear" on plain radiographs. Biopsy is not needed for diagnosis but would be expected to demonstrate replacement of mineralized bone with fibrovascular connective tissue.

Treatment appears to depend largely on the likelihood of serious morbidity and/or mortality, but spontaneous resolution requiring little intervention often occurs. This depends largely on which bones are affected; involvement of the skull, spinal cord, or chest wall can be fatal, whereas involvement of the clavicle rarely results in death but can lead to significant disability.¹² Several studies suggest surgical resection of the diseased bone and replacement with artificial bone or prosthesis as the best way to control active expanding disease.^{3,13} Replacement with artificial bone or a prosthesis has been demonstrated to be more successful than bone grafting.² Several case reports and a recent review in children indicate spontaneous resolution in many cases.¹⁴⁻¹⁶ Other treatment modalities suggested include radiation therapy, anti-osteoclastic agents, and alpha-b2 interferon.² Orthopedic consultation should be acquired when high-risk bones such as those in the thoracic wall, spine, or skull are involved or when the disease is actively expanding, but consultation is not necessary for all patients. It must be noted

that Gorham's disease has an unpredictable natural history, although many case reports tell of spontaneous resolution after 2 to 3 years.⁴ Patients with distal disease that is not expanding, such as in the clavicle or humerus, can be observed with serial radiography. In the patient presented here, conservative treatment was instituted with ongoing physical therapy as tolerated and pain control with NSAIDs as needed. His disease remained restricted to the distal clavicle without bony spread; thus, further treatment was deemed unnecessary. Pain was largely resolved and had not hindered function by the time of a follow-up encounter at approximately 8 months after injury.

Conclusion

Shoulder pain in the paraplegic population is common and manifests a vast differential diagnosis. Because SCI clinicians will likely encounter shoulder pain in their patient

population, less common clinical entities, such as disappearing bone disease, should be considered in the differential diagnosis. Currently, there is no gold standard treatment modality for Gorham's disease. Treatment options include conservative pain control with clinical and/or radiographic monitoring, surgical resection and prosthetic bone implantation, radiation therapy, anti-osteoclastic medications such as bisphosphonates, and alpha-2b interferon. Symptom severity should guide the aggressiveness of treatment. The patient should be reassured and monitored for worsening during any recovery time. In this case, Gorham's disease developed and was isolated to the right distal clavicle; it was treated conservatively with spontaneous functional resolution in less than 1 year.

Acknowledgments

The authors declare no conflict of interest.

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