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Case Report

Exploring Gorham-Strout syndrome in a young adult: an illustrative case report and literature review

Wahab Moustafa [1,‡], Jörg Silbermann [1], Salah Maksoud [2], Amr Badary [1,*,‡]

[1]Department of Neurosurgery and Spine, SRH Wald-Klinikum Gera, academic hospital of Jena University, Str. des Friedens 122, 07548 Gera, Germany
[2]Department of Neurosurgery, Darmstadt Clinical Center, Darmstadt, Germany

[*]Corresponding author. Resident Neurosurgery, Gera Clinical Center, academic hospital of Jena University, Gera, Germany. E-mail: amr.badary@hotmail.com.
[‡]Wahab Moustafa and Amr Badary authors contribute equal to the manuscript

Abstract

Gorham-Stout Syndrome (GSS) is an exceedingly rare condition characterized by bone loss and lymphatic vessel proliferation and presents diagnostic and therapeutic challenges. We present the case of a 29-year-old female with GSS manifesting as persistent headache and swelling in the left parietal bone. Initial misdiagnosis underscores the diagnostic complexity of GSS. Radiological imaging revealed characteristic features aiding in diagnosis, which were confirmed by histopathological examination showing bone tissue alterations consistent with GSS. Surgical resection and cranioplasty were successful, although post-operative complications, including late subacute hematoma and neurological symptoms, required careful management. This case underscores the importance of a multidisciplinary approach for diagnosing and managing GSS, emphasizing the importance of radiological and histopathological examinations, surgical intervention, and post- operative monitoring.

Keywords: rare bone disorder; lymphatic vessel proliferation; diagnostic challenges; surgical intervention; multidisciplinary approach; post-operative complications

Introduction

Gorham-Stout Syndrome (GSS), or vanishing bone disease, is a rare disorder marked by progressive osteolysis and lymphatic vessel proliferation. With approximately 300 reported cases globally, its incidence and prevalence remain largely unknown due to its rarity. GSS typically manifests during the second and third decades of life, although cases outside this age range have been documented [1]. While most studies suggest an equal gender prevalence, some indicate a higher occurrence in men [2].

GSS can affect any bone, presenting as monostotic or polyostotic [3]. Commonly diagnosed in children and young adults, symptoms vary by bone involvement, with localized pain, swelling, weakness, and, in severe cases, neurological defects or paralysis [4]. Commonly affected areas include the ribs, spine, pelvis, skull, collarbone, and jaw. Symptoms of GSS often include pain, swelling, and functional impairments. Clinical findings, such as osteopenia and pathological fractures, may occur and can further contribute to functional impairments [5].

Treatment options focus on symptoms management and halting disease progression. These include pharmacological approaches such as bisphosphonates, interferon alpha-2b, and sirolimus, as well as supportive procedures for complications like chylothorax [6].

This case report examines GSS in a young adult, detailing diagnostic challenges, management strategies, and postoperative care. It highlights the complexity of surgical intervention and the need for ongoing research to enhance patient outcomes.

Case presentation

A 29-year-old female patient presented with a two-year history of persistent headache and swelling of the left parietal bone area. Initially, the swelling in the left parietal bone was misdiagnosed as a lipoma, presenting as a painless, soft, mobile subcutaneous lump that was slow-growing, with no systemic symptoms. No further radiological or histopathological work-up was conducted at that time. No intervention was undertaken at that time, and the lesion was monitored for any potential symptoms that might develop. The swelling later regressed, leaving a palpable bone defect.

The patient also experienced brief episodes of unconsciousness, preceded by dizziness and hand numbness. A clinical examination revealed a conscious and cooperative patient with intact cranial nerves and no evidence of paresis. A palpable bone defect measuring 1 × 1.5 cm was noted in the left parietal bone.

The computer tomography (CT) findings included marginal sclerosis and a large defect in the left parietal skull cap, suggesting GSS (Fig. 1). Magnetic resonance imaging (MRI) confirmed a 2 cm osteolytic lesion in the left parietal bone (Fig. 2). Skeletal scintigraphy revealed moderately increased perilesional bone metabolism.

Histopathological examination of the biopsy specimen revealed compact bone tissue with fibrosis and sporadic increase in

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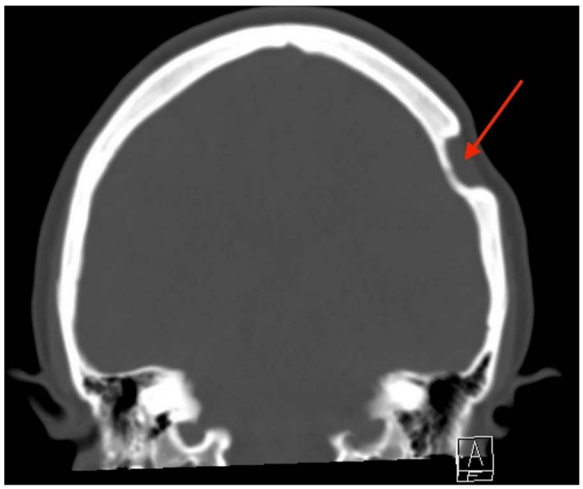

Figure 1. Coronal CT scan (pre-operative) showing a defect (arrow) in the left parietal bone.

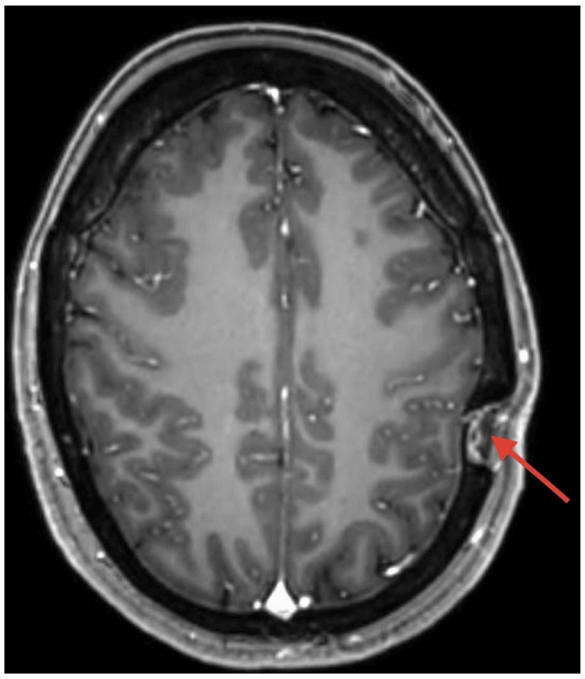

Figure 2. MRI with contrast showing lesion (arrow) in the left parietal bone.

vascularization. Foam cell accumulation, CD68 positivity, and increased vascularity were observed, consistent with GSS. Laboratory findings showed decreased erythrocyte count, hemoglobin, and hematocrit levels, as well as increased ALAT (GPT) and C-reactive protein (CRP) levels.

Surgical intervention and postoperative care

The patient underwent surgical resection of the lesion with safety margins and allogeneic cranioplasty using Palacos cement without complications. Postoperative recovery was uneventful, and proper positioning of the cranioplasty material was confirmed on the CT scan (Fig. 3).

During follow-up, MRI revealed a late subacute hematoma beneath the cranioplasty site, a known risk of cranioplasty. This highlighted the need for vigilant monitoring. An EEG showed intermittent focal slowing, leading to the initiation of levetiracetam.

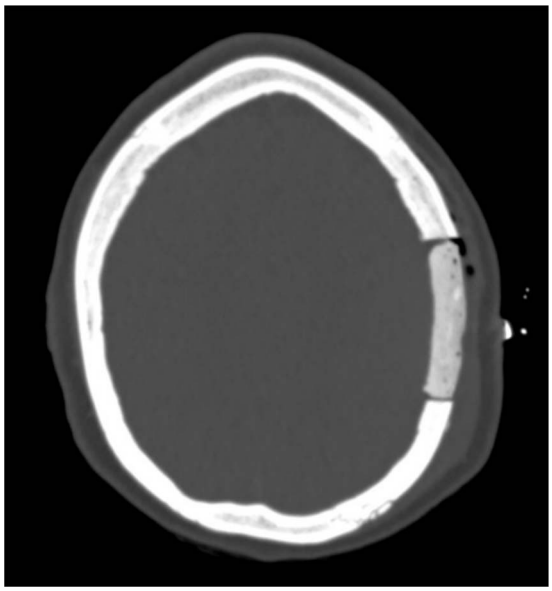

Figure 3. Sagittal CT scan (post-operative) showing proper placement of cranioplasty material (arrow).

The patient was readmitted for recurrent right leg weakness, but imaging showed no acute pathology. Conservative management resolved the weakness, and by the final examination, the patient had no significant residual neurological deficits. The patient was referred to a specialized clinic for ongoing care.

Discussion

The pathophysiology of Gorham-Stout Syndrome involves abnormal lymphatic proliferation, which plays a crucial role in the progressive osteolysis observed in affected patients. This proliferation leads to the invasion and resorption of bone tissue, contributing to the characteristic 'vanishing bone' phenomenon [7].

Misdiagnosis, as seen in our case, where GSS was initially mistaken for a lipoma, underscores the difficulty of early detection due to the nonspecific symptoms of GSS. This highlights the need for a comprehensive evaluation of unexplained bone lesions.

Radiological imaging, including CT and MRI, is pivotal in diagnosing GSS, revealing characteristic features such as marginal sclerosis and osteolytic lesions [8]. Histopathological examination confirmed the diagnosis, showing compact bone with fibrosis, increased vascularity, and foam cells positive for CD68, which are indicative of lymphatic proliferation and bone resorption [9].

To distinguish GSS from other conditions such as Giant Cell Bone Tumor (GCBT) and plasmacytoma, it's important to focus on specific radiological and histopathological characteristics. GSS is marked by diffuse osteolysis and increased lymphatic proliferation, unlike GCBT, which typically shows more defined bone destruction with multinucleated giant cells [10, 11]. Plasmacytoma, on the other hand, features monoclonal plasma cell infiltration, which was absent in this case [12].

Surgical intervention, involving resection and cranioplasty, was successful with no immediate postoperative complications. Post-operative management revealed complications, such as late-subacute hematoma, which is a known risk factor for cranioplasty. The administration of levetiracetam in response to EEG abnormalities reflects a proactive strategy for managing potential postoperative seizures, a known complication of cranioplasty and

neurological surgeries. Levetiracetam is preferred for its favorable side effect profile, ease of administration, and demonstrated effectiveness in managing focal seizure symptoms [13].

Treatment options for Gorham-Stout Syndrome (GSS) currently include pharmacological approaches such as bisphosphonates, interferon alpha-2b, and sirolimus, which target the underlying processes of osteolysis and lymphangiogenesis [14]. The patient's post-operative course, including the management of recurrent leg weakness, highlights the complex, multidisciplinary approach needed for GSS patients. The weakness was likely due to transient neurological effects or the presence of a hematoma [15].

This case sheds light on managing Gorham-Stout Syndrome (GSS) and underscores the need for further research to improve understanding and treatment. Vigilant follow-up is crucial for addressing complications promptly.

A multidisciplinary approach involving radiologists, pathologists, surgeons, and neurologists is essential. Future research should explore the molecular mechanisms of GSS, the role of lymphatic proliferation, and the effectiveness of treatments like sirolimus. Additionally, developing animal models could enhance knowledge of genetic and environmental factors affecting GSS and improve patient care.

Conclusion

A young female patient with persistent headaches and swelling in the left parietal bone was diagnosed with Gorham-Stout Syndrome (GSS) following radiological and histopathological evaluations. Successful surgical resection and cranioplasty were performed, though postoperative complications, including a late subacute hematoma and neurological symptoms, required careful management. This case highlights the importance of a multidisciplinary approach, incorporating radiological, histopathological, and surgical expertise, to optimize outcomes. Given GSS's complexity and rarity, ongoing research into its molecular mechanisms is essential for improving treatment strategies and patient care.

Human subjects

Consent was obtained from the patient for publication of the data and images anonymously.

Conflict of interest

In compliance with the ICMJE uniform disclosure form, all authors declare the following:

Payment/services info: All authors have declared that no financial support was received from any organization for the submitted work.

Financial relationships: All authors declare that they have no financial relationships at present or within the previous three years with any organization that might have an interest in the submitted work.

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