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Case Report
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Vanishing Bone Disease of the Skull Base: A Review of Rare Entity

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Abstract

Introduction: Vanishing bone diseaseis a very rare idiopathic disorder of the skeletal system of unknown etiology. The pathogenetic mechanisms of the disease remain unclear. Any part of the skeleton can be involved and the cranial localization is extremely uncommon. This case aims to highlight the diagnostic process and management of this extremely rare entity.

Case Report: A 23-year-old female, presented with asymptomatic skull defect over the left temporo-occipital region that progressively enlarged over a period of 2 years. Computed tomography revealed a left temporal osteolyticlesion, centered on the mastoidboneextending to the ipsilateral occipital bone and clivus. MRI features showed a medullary replacement process with a component whose enhancement suggests a venolymphatic vascular nature. Open bone biopsy confirmed the diagnosis of Gorham-Stout disease. We chose the conservative treatment and the results were good.

Clinical Discussion: Vanishing bone disease of the skull base is an extremely rare entity characterized by destruction of osseous matrix and proliferation of vascular structures with benign origin.The skull is among the least common locations of involvement. The diagnosis of the cranial localization of the syndrome is very challenging and it is confirmed by the histopathological analysis of the lesions. Its management is still an object of research, there is no consensus regarding the treatment of the skull localization and its prognosis is still unpredictable.

Conclusion: Gorham-Stout disease of the skull base presents unique diagnostic and therapeutic challenges. Physicians should be aware of the existence of this rare entity, early diagnosis and a multidisciplinary approach to management, including surgical and pharmacologic interventions, are crucial in optimizing outcomes.

Keywords: Gorham-Stout; Osteolysis; Vanishing bone disease; Skull base; Diagnosis; Case report

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Abbreviations

CT: Computed Tomography; MRI: Magnetic Resonance Imaging; GRAY: Gy; CSF: Cerebro-Spinal Fluid; WI: Weighted Imaging

Introduction

Vanishing bone disease also known as Gorham-Stout disease, is a very rare idiopathic disorder of the skeleton of unknown etiologyoriginallydescribed in the 1950 by Gorham and Stout [1], characterized by the uncontrolleddestructive proliferation of distendedvascular or lymphaticcapillaries withinbone [2]. Any part of the skeletal system can beaffected and the cranial localization is extremely uncommon [3]. The clinical presentation of this diseasevaries depending on the bones involved.When it concerns the skull bones, the clinical presentation includes, most frequently, pain, deformations and swelling of the affected region, although asymptomatic cases and rarely life-threatening situations mainly due to CSF leaks have been reported [3]. The diagnosis of this extremely rare entity is still challenging. There is no consensus regarding treatment and it includesthree major categories: pharmacologicaltreatment, radiation and surgery [4]. Here, we present clinical, radiological, and therapeutic data of a pathologically proved case of Gorham disease of the cranial vault.

Case Presentation

We report the case of ahealthy female with no medical history who presented at the age of 23 with an asymptomatic skull defect over the left temporo-occipital region that progressively enlarged over a period of 2 years.The defect began as a small and painless left temporal scalp depression that could not link to any previous head trauma. When the patient noted that the defect was in progress, she was referred to our institution for further evaluation.Physical examination revealed a large area about 5 x 4 cm of softening and depression over the left temporo-occipital region, there were no other abnormal findings on examination.The biological investigations were negative for malignancy, infection and immunological abnormalities. CT revealed a left temporal osteolyticlesion, centered on the mastoidbone, affecting the tympanic bone and petrous apex, extending to the ipsilateral occipital bone and the clivus. Theosteolytic areas lack surrounding sclerosis and signs of periosteal reaction (Figure 1). Cranial CT with 3D reconstruction was performed revealing an extensive bone destruction affecting both the temporal and occipital bones, particularly around the mastoid region (Figure 2). Brain MRI demonstrated continuous medullary replacement lesion, conforming to the shape of the affected bone structures, appearing as isointense on T1WI and hyperintense on T2WI with flow voids (Figure 3). Dynamic TWIST sequenceshowed a progressive lateenhancement of bonelesions (Figure 4). The preoperative differential diagnosis included juvenile Paget disease at destructive stage, eosinophilic granuloma and osteolytic metastasis but none of them correlated well with the clinical, biological and radiologic features. A left temporal open biopsy was performed. Under microscopy, the bone specimens showed intraosseous angiomatosis with mixed patterns of bone destruction, fibrous connective tissue, and abnormal small vessel proliferation (Figure 5).

On the basis of the histopathology, radiological and clinical features, Gorham's disease of the skull base was diagnosed. Immediate operative follow-up was uneventful, no complications were noted and the patient was discharged from the hospital after 2 days. Determining the most adequate treatment approach to prevent further osteolysis and protect the brain from mechanical damage was challenging since literature is not conclusive on the precise treatment approach. We decided to choose the conservative option and the treatment was initiated with bisphosphonates and radiotherapy (30–45 Gy in total). After one year, the osteolytic condition had regressed.

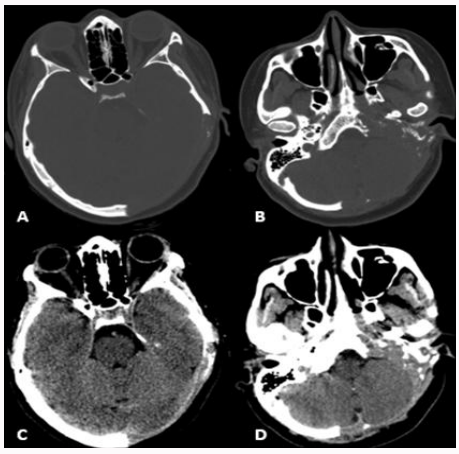

Figure 1: Axial head computed tomography (CT) bone window images (A,B) and soft tissue window images (C,D) revealing a left temporal osteolytic lesion, centered on the mastoid bone, affecting the tympanic bone and petrous apex, extending to the ipsilateral occipital bone and clivus. The osteolytic areas lack surrounding sclerosis and signs of periosteal reaction. Note the integrity of the adjacent brain parenchyma and the absence of soft tissue involvement.

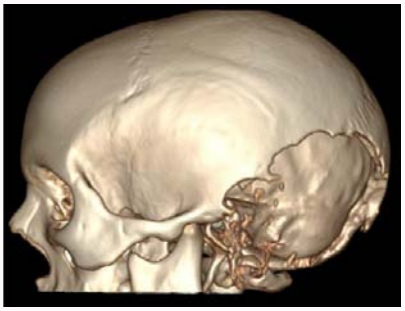

Figure 2: Head 3D volume rendering (VR) demonstrating extensive bone destruction affecting both the temporal and occipital bones, particularly around the mastoid region.

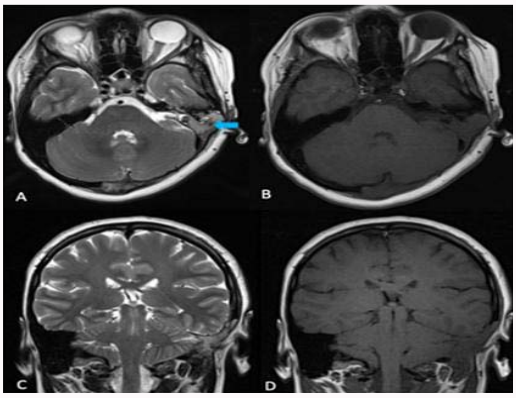

Figure 3: Brain MRI Axial T2WI (A) and T1WI (B); coronal T2WI (C) and T1WI (D) demonstrating continuous medullary replacement lesion, conforming to the shape of the affected bone structures, appearing as isointense on T1WI and hyperintense on T2WI with flow voids (A, arrow).

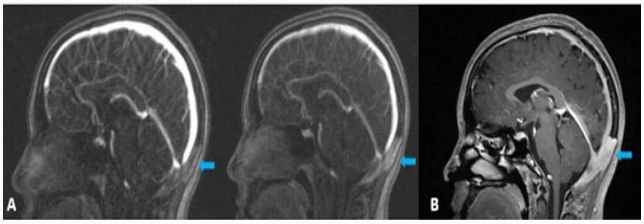

Figure 4: Dynamic TWIST sequence, sagittal images (A); Post-contrast 3D-T1-MPRAGE, Sagittal image (B) Illustrating progressive late enhancement of bone lesions (Arrow).

Discussion

Vanishing bone disease is a very rare idiopathic disorder of the skeletonof unknown etiology originally described in the 1950 by Gorham and Stout [1], characterized by the uncontrolled destructive proliferation of distended vascular or lymphatic capillaries within bone and surrounding soft tissues with benign origin [2,5]. Despite its benign character, its prognosis is unpredictable [6] and the presence of several serious complications in some cases cannot be ignored. Gorham disease is a nonhereditary disorder with no sex predilection andmost patients are younger than 40 years [6,7]. Any part of the skeletal system can be involved, especially bones that

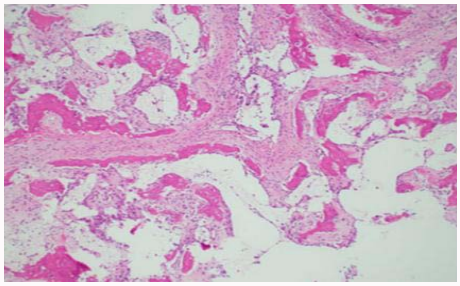

Figure 5: Intraosseous angiomatosis with mixed patterns of bone destruction, fibrous connective tissue, and abnormal small vessel proliferation.

develop by intramembranous ossification [3,8]. The skull is among the least common locations of involvement [9,11]. The exact etiology and the mechanisms of this disease remain poorly understood and it is considered by most to be due to benign, neoplastic proliferation of hemangiomatous tissue [12]. The symptoms of Gorham–Stout syndrome vary depending on the site of involvement. When it concerns the skull bones, the clinical presentation includes, most frequently, such in our case, pain, deformations, scalp atrophy and swelling of the affected region, although asymptomatic cases and rarely life-threatening situations mainly due to CSF leaks have been reported [13]. The diagnosis of the syndrome is challenging and it should be made only after excluding other etiologies of osteolysis such as immunological, infectious, malignant and metabolic affections [14]. Biological tests areusually normal. A variety of imaging methods can be used in evaluating patients suspected of having cranial vanishing bone disease. Plain radiographs and Computed tomography, initially, show radiolucent foci in the intramedullary or subcortical regions and, later, slowly progressive atrophy, dissolution, fracture and disappearance of a part of a bone, with tapering or "pointing" of the remaining osseous tissue and atrophy of soft tissues [15]. MRI is particularly useful for evaluating the extent of soft tissue involvement and vascular/lymphatic proliferation[16]. The affectedbone shows low signal intensity on T1 WI due to the loss of normal bone marrow and replacement by fibrous tissue or fluid and demonstrate high signal intensity on T2WI, indicating the presence of fluid, edema, or vascular proliferation within the affected region[16,17]. The involved regions may demonstrate enhancement due to increased vascularity or inflammatory processes within the soft tissues or fibrous replacement of bone. There may be evidence of soft tissue thickening and enhancement surrounding the areas of boneloss. No periosteal reaction or bone-forming processes are usually present, helping to distinguish it from conditions like osteomyelitis or malignancy [18]. The disease is confirmed by the histopathological analysis of the lesions [19]. Despite the fact that it is considered as begin, the cranial location of this disease has an unpredictable prognosis and possible serious complications [6,20]. There is no consensus regarding treatment and its main interest focuses on reducing symptoms to maintain health and quality of life [20]. In some cases, when the disease doesn't affect activities of daily living or cause chronic pain, the treatment may be not indicated [20].

The treatmentincludes three major categories: medicine therapy, radiation and surgery [20]. Pharmacological treatment includes mainly bisphosphonates which are thought to be beneficial through inhibition of osteoclast-mediated bone resorption and stabilization the disease process [20]. Other pharmacologic agentslike vitamin D, a-2b interferon, calcium, sirolimus, adrenal extracts and androgens have been suggested [6,20]. In most cases in the literature, such in our case, patients have been treated with monotherapy using bisphosphonates, but a combination of drugs has been used in some other cases, such as bisphosphonates with sirolimus [12] or bisphosphonates with interferon-a-2b[17]. There was no significant difference in the results. Radiation therapy with a dose of 30–40 Gy has been reported. However, radiation could provoke some serious side effects, like secondary malignancy and growth restriction in children and adolescents who receive a high-dose therapy [20]. Finally, the surgical management is performed by resection of the lesion and reconstruction by use of bone grafts and/or prostheses [20]. In the majority of cranial cases, the combination pharmacological treatment-surgeryis the most recommended [19]. If a patient has no severe deformity, CSF leak or progressive neurologic deficits, it might be better to prioritize conservative treatments and to perform the surgery after the osteolytic changes have stopped [20]. In patients with large symptomatic lesions, radiation and surgical treatment are preferred [18,20]. In this case report, we have employed combined surgical (open biopsy) and medical (Bisphosphonates and radiotherapy) management for this condition, the results were good.

Conclusion

Patients with skull Gorham-Stout disease should undergo a through clinical examination coupled with radiological, laboratory, and histopathological investigations to provide an accurate early diagnosis of this incompletely understood condition, to estimate its occurrence, and to ascertain its prognosis.Physicians should be aware of the existence of this rare entity, early diagnosis and a multidisciplinary approach to management, including surgical and pharmacologic interventions, are crucial in optimizing outcomes.

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