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Gorham-Stout disease. Case Report

Enfermedad de Gorham-Stout. Reporte de Caso

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SUMMARY

Introduction: Gorham-Stout disease (GSD) is a condition characterized by progressive bone destruction that appears between the second and third decades of life. Currently, it presents a diagnostic challenge due to its low incidence and nonspecific symptoms. There is no specific treatment for this condition, but it may include bisphosphonates, radiation therapy, and, in advanced cases, surgery.

Clinical case: A 37-year-old female patient with no relevant medical history presented with chronic intermittent pain in her left hip that had been evolving for eight months and had worsened in recent weeks, becoming constant and limiting. A simple anteroposterior (AP) X-ray of the pelvis was performed, which showed an osteolytic lesion of the right iliopubic branch, left iliopubic and ischiopubic branches, and in the pubic symphysis region. A simple computed tomography (CT) scan of the pelvis was also performed, showing an extensive lytic lesion with irregular sclerotic edges. A simple contrast-enhanced magnetic resonance imaging (MRI) scan of the pelvis suggested increased inflammatory or vascular activity in the areas described. Finally, histopathological analysis of the incisional biopsy confirmed the presence of cellular atypia and fibroangioblastic tissue without neoplastic infiltration, which are characteristic features of Gorham-Stout disease.

Conclusion: Gorham-Stout disease is a rare condition with low incidence and unknown etiology. Diagnosis requires a combination of clinical, imaging, and histopathological findings, and its treatment is currently controversial and nonspecific.

Keywords: Gorham-Stout disease, osteolysis, bisphosphonates.

RESUMEN

Introducción: la enfermedad de Gorham-Stout (EGS), es una patología caracterizada por la destrucción progresiva del tejido óseo, que aparece entre la segunda y tercera décadas de vida. Actualmente, presenta un reto diagnóstico por su baja incidencia y la sintomatología inespecífica. No existe un tratamiento específico para esta patología, pero puede incluir bifosfonatos, radioterapia y en casos avanzados, la cirugía.

Caso clínico: paciente femenina de 37 años, sin antecedentes patológicos de relevancia, presenta dolor crónico intermitente en cadera izquierda de 8 meses de evolución y que se exacerba en las últimas semanas hasta convertirse en constante y limitante. Se realizaron: radiografía antero-posterior (AP) simple de pelvis, que evidenció: lesión osteolítica de rama

iliopubiana derecha, ramas ilio e isquiopubianas izquierda y en región de sínfisis de pubis. También se realizó una tomografía axial computarizada (TAC) simple de pelvis, que mostró una lesión lítica extensa con bordes escleróticos irregulares, la resonancia magnética (RMN) simple y contrastada de pelvis sugirió actividad inflamatoria o vascular aumentadas en las zonas descritas. Finalmente, el análisis histopatológico de la biopsia incisional, confirmó la presencia de atipia celular, tejido fibroangioblástico, sin infiltración neoplásica, aspectos característicos de la enfermedad de Gorham-Stout.

Conclusión: la enfermedad de Gorham-Stout es una patología rara, con baja incidencia y etiología desconocida. El diagnóstico amerita la combinación de aspectos clínicos, imagenológicos e histopatológicos y su tratamiento actualmente es controversial e inespecífico.

Palabras clave: enfermedad de Gorham-Stout, osteólisis, bifosfonatos.

INTRODUCTION

Gorham-Stout disease (GSD), also known as idiopathic massive osteolysis or "evanescent bone," phantom bones, or lost bones1; is a rare condition characterized by progressive destruction of bone tissue (osteolysis) mediated by abnormal proliferation of lymphatic and blood vessels, leading to progressive osteolysis of the skeleton, with loss of bone mass in the affected areas[1,2]. Since its initial description by Jackson in 1838 and its characterization by Gorham and Stout in 1955, fewer than 300 cases have been documented, highlighting its diagnostic complexity over time[3-4].

Although its etiology is uncertain and unknown, several factors have been proposed, including alterations in vascular endothelial growth factor (VEGF) and the bone microenvironment, as possible triggers[4,5]. This condition can affect both children and adults, initially presenting clinically with progressive bone pain and structural weakness accompanied by pathological fractures[6-7]. Osteolysis may cease spontaneously after years of bone destruction, leading to deformity and functional impairment[7].

The diagnosis combines clinical, imaging, and histopathological findings[8-9]. Imaging tools, such as computed tomography (CT) and magnetic resonance imaging (MRI), are essential for determining the extent of the lesions, while biopsy confirms fibroangioblastic proliferation [10-11]. This pathology lacks a specific histopathological appearance but is characterized by vascular changes and the absence of cellular atypia[11].

Currently, there is no established treatment for this condition. Options include bisphosphonates, radiation therapy, and, in advanced cases, surgery. In addition, therapies with interferon alfa and denosumab have been explored with promising results, which have reported variable success[12-13]; however, none of these treatments are satisfactory when used alone. Radiotherapy is not recommended as a primary treatment and is generally used in conjunction with surgical treatment if medical therapy is ineffective[14], as this treatment has been reported to have adverse effects, such as radiation injury, when used as a single therapeutic modality, and complete resection of the lesion prevents disease progression[15].

CASE PRESENTATION

A 37-year-old female patient, of mixed race, with no relevant personal or family medical history, consulted for chronic pain in her left hip that had been present for approximately 8 months. The pain was initially intermittent until it became constant and limiting, significantly affecting her quality of life and ability to walk. Physical examination revealed pain localized in the left groin region that was exacerbated by standing and joint movement. Functional limitations of both hips were also observed, more pronounced on the left side, requiring assisted walking with a walker.

Initial laboratory tests and additional imaging were requested, including a complete blood count, blood chemistry, electrolytes, hormone tests, and other special laboratory tests, a plain anteroposterior (AP) pelvic X-ray, a plain computed tomography (CT) scan of the pelvis, and a plain contrast-enhanced magnetic resonance imaging (MRI) scan of the pelvis.

The complete blood count, blood chemistry, electrolytes, and hormone tests requested were found to be without apparent abnormalities. Special laboratory tests were also requested, including beta-2 microglobulin, whose value of 1.69 mg/L was within the normal range.

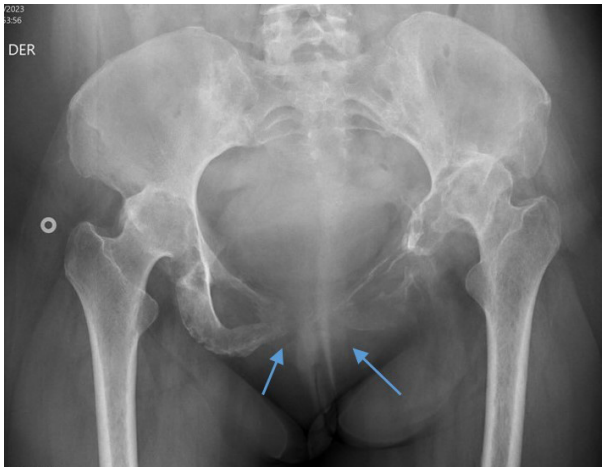

Image 1. Plain AP radiograph of the pelvis showing osteolytic lesion of the right iliopubic branch, left iliac and ischiopubic branches, and pubic symphysis region (blue arrows).

The AP pelvic X-ray revealed significant loss of bone architecture in the left ilium, with areas of severe osteolysis involving the right iliopubic branch, left iliac and ischiopubic branches, and pubic symphysis region (Image 1).

These findings were corroborated by a simple CT scan of the pelvis, which showed an extensive lytic lesion with irregular sclerotic edges, accompanied by partial collapse of the bone structure in the affected region (Image 2).

Pelvic MRI, both in simple and contrast sequences, revealed a heterogeneous signal in the affected areas, with hyperintensity in STIR sequences, located in the left hip joint, which caused dislocation of the hip joint with cephalic displacement of the humeral head and destruction of the left iliac and ischiopubic branches, without diffusion restriction, suggesting increased inflammatory or vascular activity (Image 3).

No further imaging studies were requested for this case. Based on the findings of the physical examination and the imaging studies previously requested, and considering the suspicion of an unknown pathological osteolytic process, an incisional biopsy was scheduled in the left acetabular region. The procedure was performed under general anesthesia, under strict aseptic and antiseptic conditions. The fragments obtained showed fibro-adipose and muscle tissue, with no evidence of malignancy. Additionally, no bone tissue was identified in the analyzed samples, which was consistent with advanced osteolysis in the affected area. In the absence of conclusive results and to corroborate the diagnosis, it was decided to perform a second biopsy. On this occasion, the fragments obtained included bone tissue, soft tissue, and muscle tissue. Histopathological analysis confirmed the presence of fibro-angioblastic tissue, without neoplastic infiltration, significant proliferation of lymphatic and vascular channels with cellular atypia, findings characteristic of Gorham-Stout disease. (Image 4).

This result, together with the clinical and imaging data described above, allowed us to establish a definitive diagnosis of the pathology. After diagnosis, conservative treatment was initiated immediately, consisting of oral ibandronic acid (150 mg monthly), calcium supplements (500 mg daily), and vitamin D (5,000 IU daily). During subsequent follow-up visits, the patient reported progressive clinical improvement, with decreased pain and partial recovery of function, although she continued to require a walker for ambulation. Follow-up radiographic studies showed stable progression of osteolysis (Image 5).

The patient's symptoms gradually decreased, especially pain, which was corroborated by periodic clinical and im imaging examinations. In this context, one year after starting conservative treatment, the patient's functional status is acceptable, with stabilization of the disease, and she continues to undergo medical check-ups every six months.

DISCUSSION

Gorham-Stout disease (GSD) is a rare disease, with fewer than 300 cases reported in the medical literature to date. It occurs in both men and women, usually in the second and third decades of life[1,14], although cases outside this age range have been reported. It affects any bone structure, but is most common in the ribs, vertebrae, and pelvis. In the upper extremities, the most affected bone is the humerus, and in the lower extremities, the femur and tibia[2,15]. The clinical presentation typically begins with fractures and severe pain, accompanied by a range of symptoms that vary depending on the affected bone[2-3]; therefore, it presents both a diagnostic and therapeutic challenge due to its low incidence and nonspecific clinical presentation[3]. The rarity of this pathology, together with the clinical presentation characterized by progressive bone destruction in the pelvis, corroborated by imaging

studies and biopsy, are essential parameters for a specific therapeutic approach[2]; it should be noted that osteolysis in this pathology can be estimated in complementary radiographic studies, while the diagnosis of SGE can be confirmed with various histopathological studies[3]. However, it should be emphasized that in patients with skeletal osteolysis, a thorough medical history and physical examination, including blood tests and radiographic studies, should first be performed to rule out other underlying diseases such as infection, cancer (primary or metastatic), inflammatory or endocrine disorders, chronic osteomyelitis, and Paget's disease[4].In this context, the absence of systemic symptoms, such as fever, and a history of cancer, together with a specific physical examination, specific imaging findings, and the histopathology report, allowed these possibilities to be ruled out in the patient in question[5].

Imaging tools, especially computed tomography (CT) and magnetic resonance imaging (MRI), were crucial for assessing the extent of bone involvement[6]. CT allowed the lytic lesions and their sclerotic edges to be defined. At the same time, hyperintensity on MRI STIR sequences indicated inflammatory and fibroangioblastic activity, allowing for differentiation between the early, active, and late stages by demonstrating changes in signal intensity over time, resulting from inflammation, increased capillary permeability, and fibrosis. Histopathologically, it is characterized by intraosseous vascular endothelial proliferation leading to focal bone resorption, fibrosis, replacement, fracture, and changes in adjacent soft tissue, as well as the absence of cellular atypia[7,15].

As for therapeutic management, current evidence does not recommend any specific treatment. However, different modalities have been tried, ranging from conservative drug treatment to surgical resection techniques, including radiotherapy[8,9,15]. Pharmacological treatment includes octreotide, calcitonin, bevacizumab (an anti-VEGF-A monoclonal antibody), propranolol, interferon alfa-2b, sirolimus, low-molecular-weight heparin, and bisphosphonates, especially third-generation bisphosphonates[9,16]. In our patient, treatment with bisphosphonates (ibandronic acid) was used in conjunction with calcium and vitamin D to stabilize the osteolytic process, yielding good clinical results. Bisphosphonates are effective in inhibiting osteoclastic activity, although the available data are primarily based on case reports and observational studies[10-11]. In more advanced or refractory cases, options such as radiotherapy have been considered to halt disease progression[12-13]. In this context, there is no definitive treatment for this condition, as many of the treatments mentioned above are still under investigation[13-15].

Despite the rarity of EGS, this case highlights the importance of a multidisciplinary approach involving orthopedics, radiology, and pathology[13]. Although

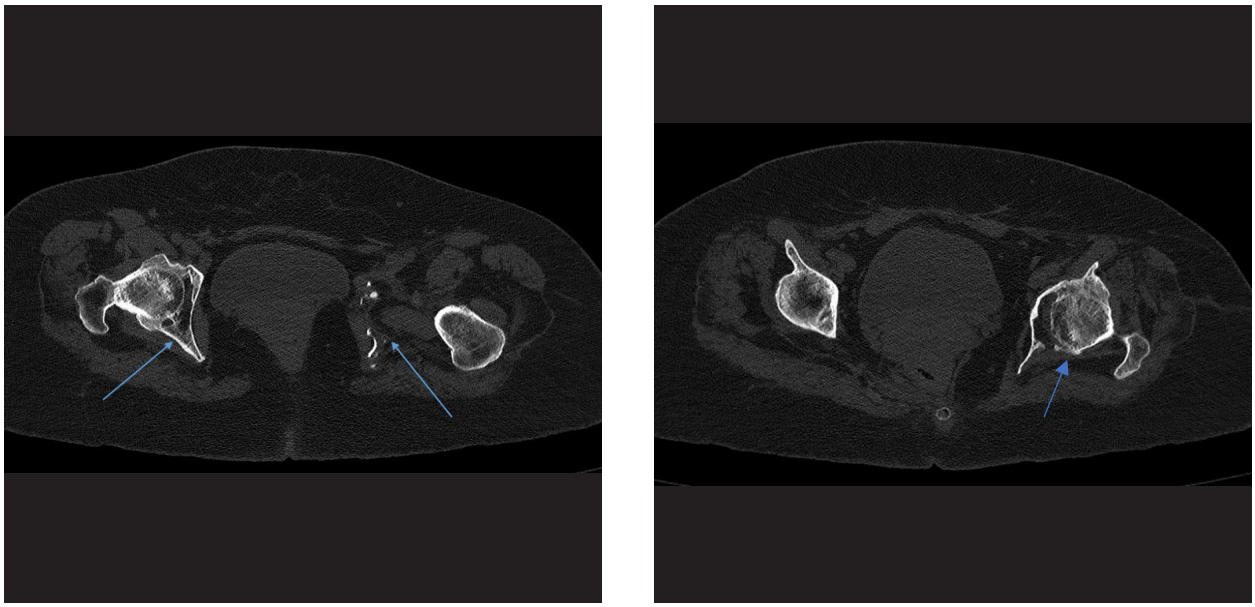

Image 2. Pelvic CT scan showing the extent and characteristics of the extensive osteolytic lesion in the left iliopubic and iliac branches, with irregular edges and areas of peripheral sclerosis (blue arrows).

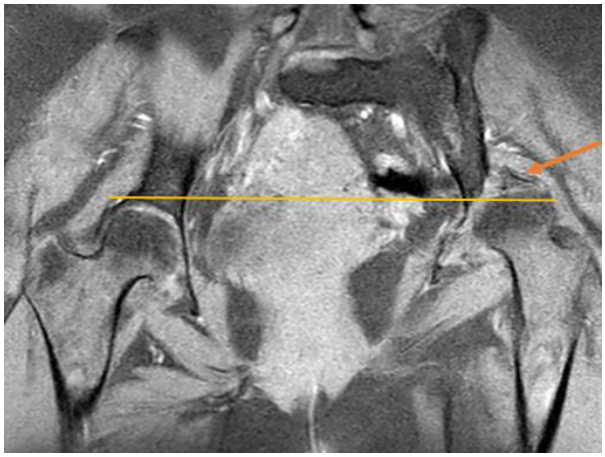

Image 3. MRI of the pelvis, coronal section showing cephalic displacement of the humeral head (tomato arrow).

treatment stabilized the patient's condition and improved her quality of life, continuous follow-up is essential to identify possible complications or disease progression[14,15]. The prognosis varies significantly in each patient, depending on the affected area and the disease's stability. Therefore, complications depend on the location and extent of its presentation. The disease may be self-limiting or lead to death; there are reports of patients who experience spontaneous remission and others in whom the disease progresses so aggressively that it is life-threatening within a year[15]. Regarding the case reported in our patient, the literature reports that involvement of the lower extremities or pelvic girdle has a good prognosis[16].

CONCLUSSION

Gorham-Stout disease is a rare condition that poses a significant diagnostic and therapeutic challenge due to its low incidence and nonspecific clinical presentation. Imaging studies, particularly CT and MRI, are crucial for evaluating the extent of the disease and monitoring its progression. Conservative management, based on bisphosphonates, calcium supplements, and vitamin D, is effective in stabilizing disease progression and improving patients' quality of life. However, continuous clinical and imaging follow-up is required to evaluate response to treatment and detect possible complications in the short, medium, and long term.

BIOETHICAL ASPECTS

This clinical case has been approved by the patient, and written informed consent has been obtained. The confidentiality and anonymity of the patient have been guaranteed at all stages of the report.

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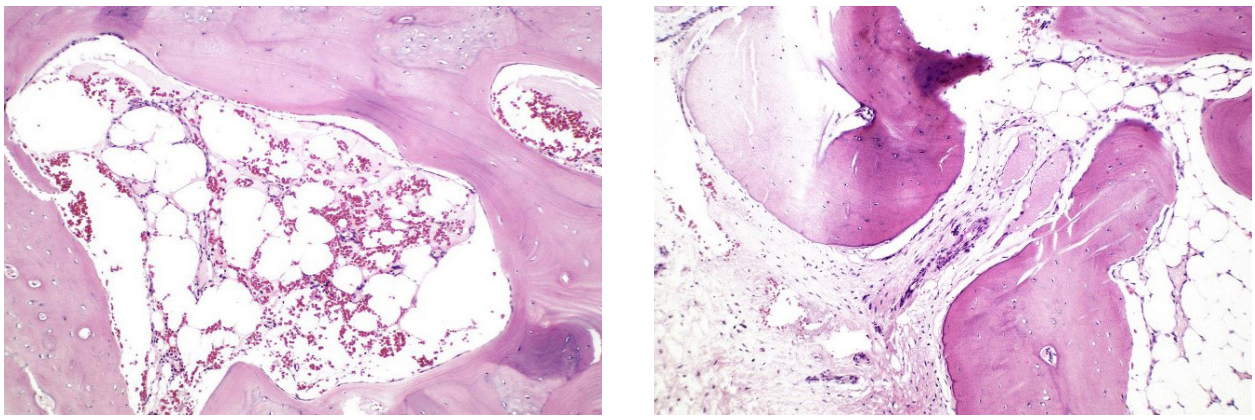

Image 4. Microscopic examination, with hematoxylin and eosin staining, reveals cellular atypia characterized by thin bone trabeculae and intermediate spaces, accompanied by significant proliferation of lymphatic and vascular channels.

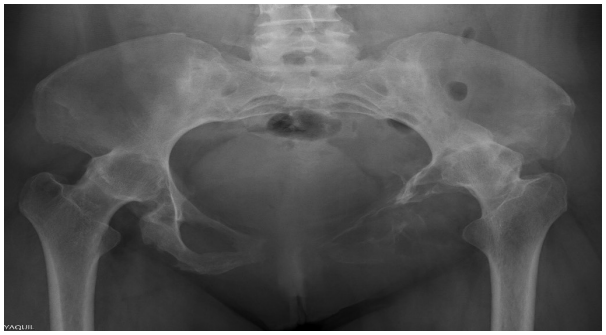

Image 5. Follow-up pelvic X-ray showing stabilization of the osteolytic process after the start of treatment.

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AUTHORS' CONTRIBUTIONS

M.F.B.C: Participated in the conception and design of the work, as well as in the analysis and interpretation of the data. Contributed to the writing and critical review of the manuscript, approved the final version, and assumed responsibility for all aspects of the article.

J.M.J: Participated in the conception and design of the work, including the analysis and interpretation of the data, and approved the final version.

I.V.S: Collaborated in the conception and design of the work, also participated in the writing and critical review of the manuscript, approved the final version, and ensured responsibility for all aspects of the article.

K.A.D: Participated in the conception and design of the work, including the analysis and interpretation of the data, and approved the final version.

CONFLICT OF INTERESTS

The authors declare that there is no conflict of interest.

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