Gorham disease. A case report

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Background:
Gorham-Stout Syndrome (GSS), also known as vanishing bone disease or massive osteolysis, is a rare disorder characterized by the spontaneous and progressive resorption of bone tissue. The cause of this syndrome remains unknown, and its unpredictability and rarity pose significant challenges for diagnosis and treatment. This article explores the clinical manifestations, diagnostic procedures, and current treatment approaches for Gorham-Stout Syndrome.

Keywords: Gorham-Strout Syndrome, vanishing bone disease, massive osteolysis.

Quetzaltenango, Guatemala

Case Report

Radiology

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Gorham disease, also known as Gorham-Stout disease or vanishing bone disease, is a poorly understood rare skeletal condition which manifests with massive progressive osteolysis along with a proliferation of thin walled vascular channels. The disease starts in one bone but may spread to involve adjacent bony and soft tissue structures.

Case report

18-year-old male patient, originally from Momostenango, Totonicapán, occupation of merchant. Patient reports that approximately 10 years ago he suffered a fall from a tree causing a fracture of the right radius, he consulted this health center where he received surgical treatment; however, he did not return for follow-up and the patient reports that over the years the arm presented a certain degree of deformity and approximately 1 year ago he noticed that the forearm began to present shortening, associated with which he reports moderate pain, partial loss of mobility and sensitivity.

Biopsy results showed chronic osteolysis, osteosclerosis and chronic xanthomatous osteomyelitis. Based on clinical and radiological findings, which show spontaneous and progressive bone resorption, Gorham-Stout Syndrome is diagnosed.

Discussion

Pathophysiology of the lesion

It presents as an idiopathic osteolysis, which can affect any bone, but most frequently the scapula and the mandible, and can spread to adjacent bones and even cause the affected bone to disappear completely.

Gorham and Stout described the main pathological characteristics of this rare disease as non-malignant intraosseous proliferation of vascular tissue that causes malignant osteolysis.

There are three fundamental characteristics in the etiopathology of SGS, which are the role of osteoclasts, angiogenesis/lymphangiogenesis and the function of osteoblasts.

Clinical manifestations and diagnosis

They are variable and depend on the site of the condition, and generally follow a chronic and progressive course.

The diagnostic criteria are as follows:

1. Absence of cellular atypia.
2. Minimal or no osteoblastic response and absence of

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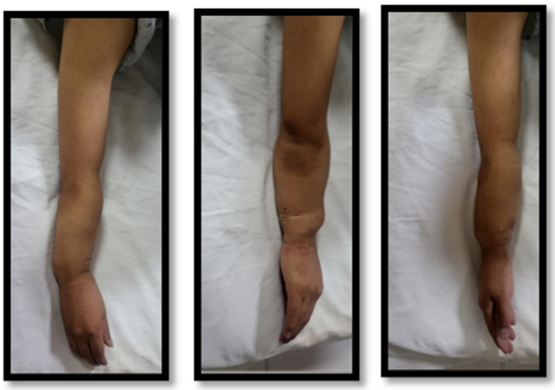

Figure 1. Illustrative photos with the patient's permission.

dystrophic calcification.
3. Evidence of progressive bone resorption.
4. Non-expansive and non-ulcerative lesions.
5. Absence of visceral involvement.
6. Radiographic osteolytic pattern.
7. Without hereditary, metabolic, immunological or infectious cause.

Imaging studies

Radiographically, 4 stages are distinguished:

First: It is an intraosseous stage characterized by multiple medullary and subcortical lucencies that resemble osteoporosis.

Second: There is an increase in these radiolucent foci that tend to coalesce, having poorly defined margins.

Third: It is characterized by cortical erosion and local invasion of the angiomatous mass into the adjacent soft tissue.

Fourth: There is complete resorption of the affected bones with subsequent replacement by fibrous tissue.

The findings in tomography will be similar, and can show the extension into the surrounding soft tissues.

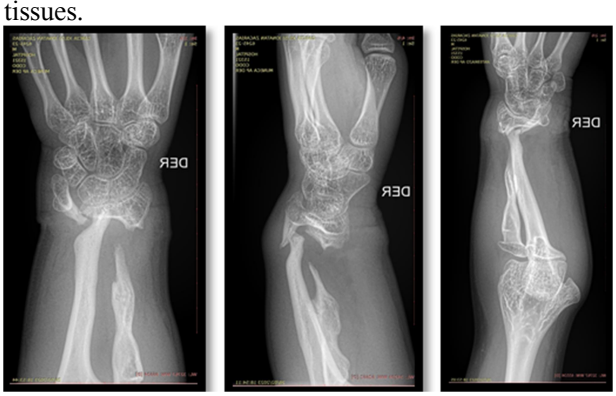

Figure 2. X-ray of the right wrist and forearm. Distortion of the anatomical configuration of the ulna and distal radius is observed, which present bone resorption, being more evident at the level of the distal third of the ulna, which causes the radioulnar joint to not

Figure 2. Contd. visualized. There is sclerosis at the level of the radiocarpal joint, as well as marked bone demineralization of the carpal bones and the distal end of the radius.
Proximally, the ulna and radius are also visualized, which are observed with an alteration of their normal configuration

Treatment

Various methods have been used, with limited results. Surgical interventions for bone implantation have been used with poor results. Radiotherapy with moderate doses and anti-osteoclastic and anti-hemangiomatosis medical treatment have also been used.

Conclusion

It is a rare disease in which spontaneous and progressive bone resorption occurs. This syndrome is also known by different names: massive osteolysis, vanishing bone disease or phantom bone. It presents as an idiopathic osteolysis, which can affect any bone, but most frequently the scapula and the jaw, and can spread to adjacent bones and even cause the affected bone to disappear completely.

Conflicts of interests

There was no conflict of interest during the study, and it was not funded by any organization.

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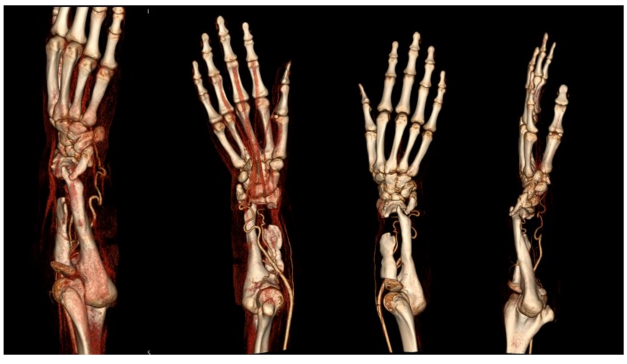

Figure 3. We can see a 3D reconstruction showing the findings described above in the X-ray. However, in this angiographic phase, tortuosity is observed, especially in the ulnar and interosseous arteries and to a much lesser degree in the right radial artery.

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