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VANISHING BONES – GORHAM IDIOPATHIC OSTEOLYSIS. A RARE DISEASE.

Radiology

Sultan Nawahirsha* Department of General Medicine, Christian Medical College, Vellore, Tamil Nadu, India *Corresponding Author

David Mathew Department of Radiology, Christian Medical College, Vellore, Tamil Nadu, India

Tahiri putta Department of Radiology, Christian Medical College, Vellore, Tamil Nadu, India

Ramya I Department of General Medicine, Christian Medical College, Vellore, Tamil Nadu, India

ABSTRACT

Gorham's disease is a rare osteolytic disease of unknown etiology. It is characterized by massive osteolytic resorption of the bone which is replaced by lymphangiomatosis of the bone(1). It is also known as Gorham Stout disease, disappearing bone disease and idiopathic osteolysis.(2)The diagnosis is established with bone biopsy and evidence of progressive osseous resorption in imaging (3). Here, we report a case of a 27 year old male who presented with chronic holo-cranial headache and bilateral sensorineuronal hearing loss. On investigations, he was found to have multiple well circumscribed lytic lesions and his biopsy revealed benign angiomatous lesion thus fulfilling the diagnostic criteria proposed by heffez and colleagues in 1983. He was managed with bisphosphonates.

KEYWORDS

INTRODUCTION:

Gorham Stout disease is a rare entity of unknown etiology characterized by osteolytic resorption of the bone which is replaced by lymphangiomatosis of the bone(1). Gorham's disease has varied clinical presentation, ie it can involve the cranium, mandible, spine.(4) It is also known as Gorham stout disease, essential osteolysis, disappearing bone disease , idiopathic osteolysis, phantom bone disease(3). In 1983 jackson, hardegger et al ,reported a case of idiopathic osteolysis characterized by rapid bone resorption of the involved bone(5).

CASE REPORT:

A 27 year gentle man from South India, without any addictions or medical comorbidities presented to us with complaints of chronic, mild, intermittent headache for 14 years with a recent worsening of severity and persistent, bi frontal, throbbing type of head ache of 1 month duration, with no history of obvious aggravating and relieving factors. There was no history of fever, projectile vomiting, diplopia, gait disturbances, motor weakness or sensory deficits.

On general examination, he was comfortable, conscious, oriented and had no dysmorphic facies. He was afebrile and his pulse rate was 84/min and blood pressure was 120/80 mmHg. He had a mild swelling of his right zygomatic bone. He was found to have Rinnes normal bilaterally, with Weber's lateralized to the right ear. His fundus was normal and the remaining Central nervous system examination was unremarkable. Other systems examinations were normal.

For the above clinical picture he was suspected to have chronic meningitis elsewhere and was advised a brain imaging. Computed Tomography [CT] of the brain had shown multiple lytic lesions over the skull. At this point, he was started on empirical anti-tuberculous treatment and was referred for further management. In view of the chronic headache, a repeat CNS imaging was done, which ruled out any features of meningeal inflammation or raised intracranial pressure. Instead, it confirmed the presence of an osteolytic lesion in the base of the skull, multiple geographic lytic lesions noted in the calvarial bones, sphenoid bone, right maxilla, zygomatic arch and the right mandibular condyle [Figure-1A]. X ray of the hip revealed similar osteolytic lesion [figure 1-B]. Audiometry done showed bilateral sensorineural deafness, with the left side being more severe.

In view of the history of an intermittent headache with sensorineural loss and multiple lytic lesions over the skull, and absence of raised intracranial pressure and features of meningeal inflammation the differentials considered were Pagets' disease, HIV with opportunistic mycoses, Langerhans cell histiocytosis , Multifocal castleman's disease, Hyperparathyroidism, malignancy with secondary metastasis All the following differentials were ruled out by appropriate investigations as shown in [table 1]

Table 1. – Relevant Investigations

	Reference range	Patient's values
Hematological investigations		
Hemoglobin (g/dl)	13.3-16.2	15.2
Total leukocyte count (mm3)	4000-12,000	8100
Platelet count (lakhs/mm3)	150,000-400,000	2,09,000
Prothrombin time (s)/international normalized ratio	10-12.5	1.01
Activated partial thromboplastin time (s)	23.5-35.5	37.3
Biochemical investigations		
Creatinine (mg%)	0.5-1.4	0.55
Sodium (mmol/L)	135-145	138
Potassium (mmol/L)	3.5-5.0	4.4
Bicarbonate[mmol/L]	22-29	26
Serum Alkaline phosphatase [U/L]	40-125	84
Total protein [g/dl]	6.0 – 8.5	6.7
Serum albumin [g/dl]	3.5-4.5	4.3
Urine bence johnes		negative
Calcium [mg%]	8.3-10.4	8.73
Phosphorous [mg%]	2.5-4.6	3.2
ParaThyroid Hormone	8.0-74	74
Vitamin D [25OH]	>30	12.1
Serum Electrophoresis		Normal
Microbiological Investigations		
HIV 1 and 2 antibodies [HIV]		Negative
Hepatitis B surface antigen [HBSAG]		Negative
Hepatitis C antibody [HCV]		Negative
Bone Marrow		Cellular marrow with much intramedullary hemorrhage and trilineage haematopoiesis , trephine biopsy

Bone Biopsy = Right Zygoma		Benign angiomatous lesion, biopsy right zygoma

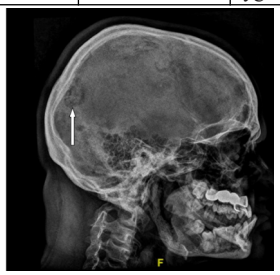

Figure 1A

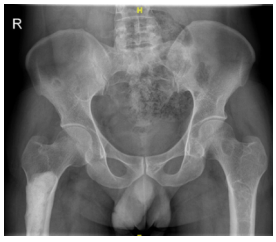

Figure 1B

PET CT [POSITRON EMISSION TOMOGRAPHY] was done which ruled out the presence of any visceral malignancy, generalized lymphadenopathy or increased uptake. [Figure 2A , 2B , 3A ,3B]

A PET CT done showed numerous, well-circumscribed, lytic lesions throughout the skeleton including the skull and facial bones. [Image 2A] Many of the spine lesions revealed a thin sclerotic rim along with prominent trabeculae around. [Image 2B]Right maxilla, zygomatic bone and right ramus of mandible also had similar findings [Image 3A, 3B]. The morphology of these lesions were in favor of hemangiomas and lymphangiomas. There is no evidence of abnormal or increased metabolic activity in any of these bone lesions.

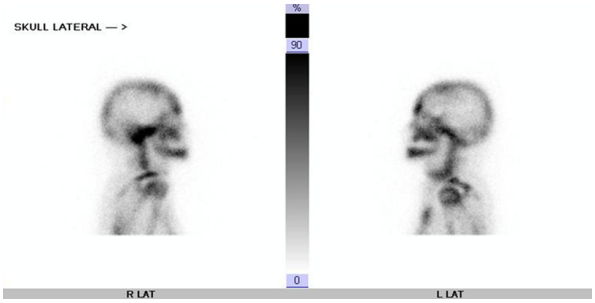

Figure 2A **Figure 2B**

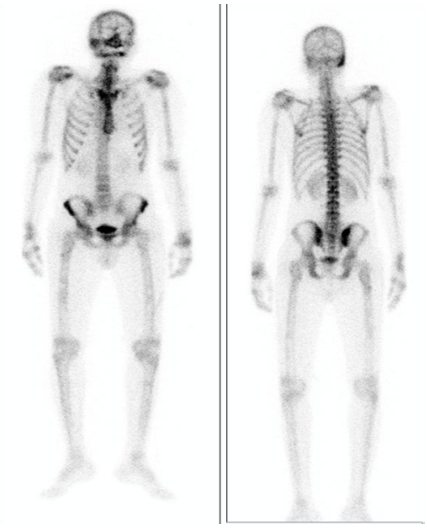

Figure 3A **Figure 3B**

Biopsy done from the right zygomatic bone showed trabeculae of cortical and cancellous bone with intervening marrow completely replaced by dilated cystic spaces lined by flattened endothelium and filled with blood. Evidence of abundant reactive new bone formation with sclerosis was present. The stroma showed areas of fibrosis and hyalinization. The above histopathological findings were diagnostic of benign angiomatous lesion, biopsy, right zygoma. At this point in time the clinical, radiological, histopathological features were diagnostic of Gorham's disease. He was started on IV Zolendronic acid 4mg bisphosphonates (7) and vitamin D supplementation [inj Arachitol 6 lakh units IM]. After initiation of treatment, he was discharged in a stable condition. At the end of one month, the patient had come to the outpatient department and was found to be doing better.

DISCUSSION:

Bone destruction where a meticulous search for etiology like infection, trauma, neoplasia or metabolic syndromes fails to reveal an etiology is termed as Essential osteolysis. Graham stout disease is a rare disorder of idiopathic massive osteolysis of unknown etiology. Less than 200 cases have been reported worldwide. It is also known as massive osteolysis ,disappearing bone disease , vanishing bone disease , phantom bone disease (3). It is characterized by Replacement of normal bone by an aggressively expanding but non-neoplastic vascular tissue (8). 2 cases of disappearing bone disease was reported by Graham et all in 1954, and bone biopsy and histological findings have shown the proliferation of thin walled , capillaries within the vanishing bone with evidence of angiomatosis.(9) Gorham had hypothesized that trauma initiated local vascular granulation tissue that brought about osteolysis by hyperemia, change in the pH, and mechanical forces (9).Even though the disease has been known since 1900 , the etiology of the disease isn't clear. Many hypotheses have been made to explain the etiology based on the histology findings of the disease. A study conducted by delving et al, had shown the increasing proliferation of osteoclast-like multinucleated cells in the serum of patients affected the disease. Elevated levels of interleukin-6 (IL-6) have also been shown to be present in this condition .(10) Histologically, involved bones show a non-malignant proliferation of thin-walled vessels; the proliferative vessels may be capillary, sinusoidal or cavernous. In late stages, there is progressive dissolution of the bone leading to massive osteolysis, with the osseous tissue being replaced by fibrous tissue.(10) Moller G et al , had described a case series involving 6 patients have found evidence to suggest that increased osteoclastic activity results in the resorption of the bone.(11) Depending on the bone involved, the patients can present with a wide variety of symptoms. 30 % of patients present with maxillofacial involvement. Patients usually seek the dentist for un resolving toothache and jaw pain due to the involvement of the mandibular bone.—(4) Patients also present to the orthopaedician with the complaints of chronic progressive back pain, not relieved with analgesics. History of pathological fractures involving the cervical spine and deformity of the spine is also a known presentation of the disease. patients can also present with complaints of a chronic headache similar to our patient along with signs of raised intracranial pressure like projectile vomiting and papilledema (2). Halliday et al, presented a case of paraplegia in a young male, who was later diagnosed to have lymphangioma of the bone. Due to the varied clinical presentation of this rare disease, it can be misdiagnosed and unreported. A high clinical suspicion is needed to diagnose this rare disease. In view of the atypical features Heffeze et al., proposed the diagnostic criteria for establishing the diagnosis of massive osteolysis as:(6) Which could be further subgrouped into skeletal and pathological criteria along with two exclusion criterias.

Skeletal criteria:

- Minimal or no osteoblastic response and absence of dystrophic calcification
- Evidence of local progressive osseous resorption
- Non- expansile, non-ulcerative lesion
- Osteolytic radiographic pattern

Pathological criteria:

- A positive biopsy for angiomatous tissue
- The absence of cellular atypia

Exclusion criteria:

- The absence of visceral involvement
- Negative hereditary, metabolic, neoplastic, immunologic or infectious etiology.

Our patient fulfilled the diagnostic criteria as proposed by Heffeze et al.

There is no definite treatment for this disease. Many case reports have shown some benefit by using bisphosphonates in decreasing the osteoclastic activity of this disease.(12)

Radiation therapy has also been shown to improve the clinical outcomes of some patients.(13)

Many case reports have published promising results when surgical intervention and radiation therapy was combined.(14)

Few case reports have shown some benefit in delaying the disease progression of patients with alpha 2b interferon.(12)

CONCLUSION

Gorham stout disease is a rare idiopathic massive osteolysis with diagnostic challenges. Multiple osteolytic lesions in the absence of systemic involvement should prompt the clinician to suspect the same. Since the prognosis is varied, Heffeze's diagnostic criteria including clinical, radiological and pathological diagnosis are instrumental in establishing the early diagnosis and treatment of the patient.

CONFLICT OF INTEREST STATEMENT

The authors declared no potential conflicts of interest with respect to the research, authorship and/or publication of this article.

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