GAVIN PUBLISHERS

Journal of Orthopedic Research and Therapy

Jakubietz MG, et al. J Orthop Ther: JORT-147.

DOI: 10.29011/2575-8241.000047

Case Report

Gorham Stout Syndrome Presenting with Vascular Symptoms of The Skin

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Citation: Jakubietz MG, Jakubietz RG, Gruenert JG (2017) Gorham Stout Syndrome Presenting with Vascular Symptoms of The Skin. J Orthop Ther: JORT-147. DOI: 10.29011/2575-8241.000047

Received Date: 11 August, 2017; **Accepted Date:** 12 December, 2017; **Published Date:** 19 December, 2017

Case

The Gorham Stout Syndrome is a rare osteolytic condition. It can occur at any age, but predominantly can be found in children and teenagers. Diagnosis is established considering clinical appearance, radiographic studies and histological results. An occurrence of Gorham Stout syndrome is presented, where osteolysis was preceded by vascular symptoms of the skin. Osteolysis was confined to the distal phalanx of the little finger and required no surgical intervention. The osteolysis eventually came to a halt, but vascular symptoms recurred episodically. This presentation shows that in recurrent vascular lesions in a child, a Gorham Stout syndrome should be taken into consideration when all other diagnoses have been ruled out.

The Gorham Stout Syndrome is a rare, massive idiopathic osteolysis [1,2]. It presents as a spontaneous, monocentric and progressive bony resorption without being confined to boundaries such as joints. It is commonly seen in young patients, although it has been reported at any age. The majority of cases occur in the skull, shoulder, pelvis and carpal bones. The etiology is unknown, a genetic predisposition has not been proven in the literature. A spontaneous arrest of osteolysis is typical, while to this date no established therapy exists. The Gorham Stout syndrome, also known as vanishing bone disease, phantom bone or acute spontaneous absorption of bone, was first described more than 150 years ago [1-3]. Fewer than 200 cases have been reported in the literature. The initial manifestation is usually a spontaneous, pathologic fracture with subsequent absence of bony regeneration. Radiological findings are progressing radiolucent foci in the intramedullary and subcortical areas resembling patchy osteoporosis [4,5]. The bone vanishes over time, shrinking in a concentric fashion, often described as 'sucked candy'. Osteolysis in the upper extremity usually comes to a spontaneous arrest while lethal complications rarely occur when only one extremity is involved. No generalized symptoms are present despite localized pain, swelling and progressive deformity. Surprisingly good function of the involved extremity is usually encountered despite a significant deformity.

No Gorham Stout Syndrome in combination with vascular changes of the skin has been reported to our knowledge. An occurrence of unilateral involvement of only the right little finger in a 10-year-old girl was seen. The initial symptom was a sudden-onset extensor lag of the distal interphalangeal joint without any history of previous trauma. No therapeutic or diagnostic interventions were initiated. Three years later a subdermal, patchy, confluent hematoma, involving the complete digit, developed spontaneously in the same finger which was tender to palpation. This perivascular exudate like symptom resolved within 2 days. Radiographic studies then revealed disappearing bone and osteolysis, which only involved the distal phalanx (Figure 1).

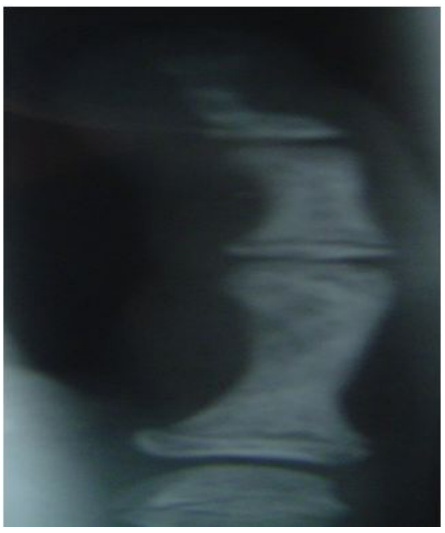
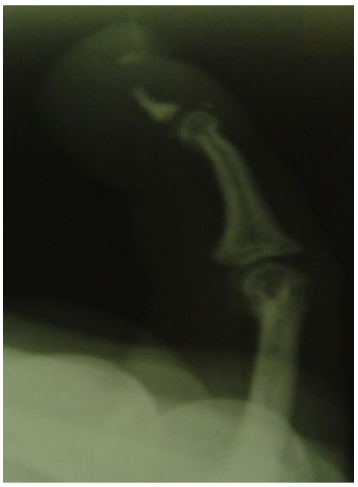


Figure 1: Osteolysis of the distal phalanx.

An angiography, intended to rule out systemic vascular disease, showed rarefied digital vessels only in the involved digit. Histologically, bony resorption, enlarged osteoclasts and granulation tissue with newly formed capillary vessels were confirmed. The disease progressed over the next 5 years, with several radiographic studies showing progressive osteolysis (Figure 2).



Hemangiomatosis in combination with osteolysis as well as the radiographic and clinical results established the diagnosis of Gorham Stout syndrome. Osteolysis came to a halt at the age of 18 and showed no progress ever since. The DIP joint has been respected as a boundary. The distal phalanx had shortened slowly to approximately 50% of the initial length (Figure 3).

Figure 3: Final appearance.

Except for a persisting extensor lag of the distal phalanx and a minor deformity no other impairment is obvious. Nevertheless, the patient had 4 similar recurrent vascular episodes which always resolved within a few days. Biopsy during an episode revealed perivascular exudates, while infection was ruled out.

The diagnosis of Gorham Stout syndrome can be reached by exclusion of other diagnoses, for which a systematic approach is recommended. First, an infectious cause has to be excluded. Osteolysis in a child may be due to osteomyelitis, either by direct inoculation or hematogenous spread. Leukocytosis, elevated CRP and positive cultures lead to this diagnosis, besides radiographs. Infections with herpes simplex virus need to be ruled out as well. Radiographs also can exclude melorheostosis, where, in contrast to osteolysis, excessive bone deposition is visible, despite a clinically similar picture. Malignant tumors and metastases can also cause osteolysis. Osteolysis as a late complication of cold exposure can be excluded by patient history. Collagen vascular disorders such as systemic lupus erythematodes or scleroderma can be ruled out by immunologic studies and skin biopsies. Also, endocrine and metabolic disorders such as hyperparathyroidism, although not commonly showing localized osteolysis need to be excluded. The Winchester syndrome, showing a similar clinical picture is inherited in an autosomal recessive pattern and also needs to be excluded.

The Gorham Stout Syndrome is a rare osteolytic condition. It can occur at any age, but predominantly can be found in children and teenagers. Diagnosis is established considering clinical appearance, radiographic studies and histological results such as intraosseous capillary proliferation, angiomatosis and fibrosis in later stages. In 20% of the patients a clear histological diagnosis cannot be established [6]. The pathogenetic mechanism is controversial. Angiomatosis and osteoclastic activity are thought to be responsible [7]. Additional factors are dysplasia of sympathic blood vessels, local hypoxia, acidosis and neurovascular irritation. The osteolytic deadspace is filled up with fibrous tissue. Other explanations include activation of previously silent hamartoma or a primary aberration of vascular tissue in the bone [8,9]. Neurovascular changes similar to the Sudeck dystrophy have also been described [10]. More recent reports describe hyperactive osteoclasts [1]. Until now, no reports have described additional symptoms involving the soft tissue and the skin. In this patient the only symptom for an extended period of time were transient vascular changes in the skin. Only after a progressive shortening of the distal phalanx radiographic and histological findings confirmed the diagnosis. Osteolysis eventually came to a halt, but vascular symptoms recurred episodically. These symptoms were treated with steroids although a spontaneous regression had always occurred. No other medication has been taken by the patient. No established medication and other proven treatment options exist and although antiresorptive treatment such as biphosphonates or calcitonin have been suggested [7]. As for now, therapy can only alleviate the symptoms and since operative intervention cannot cure the disease, such approaches should be confined to the most severe cases.

Gorham Stout syndrome has not been associated with vascular symptoms in the skin so far. In this patient cutaneous signs were confined to a single digit and were initially the most obvious

symptom. The Gorham Stout Syndrome is a rare osteolytic condition. It can occur at any age, but predominantly can be found in children and teenagers. Diagnosis is established considering clinical appearance, radiographic studies and histological results. The osteolytic process only involved the distal phalanx and did not cross the joint unlike in typical cases of Gorham Stout syndrome. This case shows that in recurrent vascular lesions in a child in combination with osteolysis, a Gorham Stout syndrome should be taken into consideration after all other diagnoses have been ruled out.

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