‘Vanishing carpal bones’ in a patient with congenital adrenal hyperplasia – A diagnostic dilemma

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ARTICLE INFO

Keywords:

Gorham's disease
Vanishing carpal bones
Wrist

ABSTRACT

The vanishing bone disease, also known as Gorham's disease usually involves long bones. Isolated carpal bone involvement is uncommon. A 14 year old male presented with pain in the right wrist joint since 4 months. He was a known case of Congenital Adrenal Hyperplasia on oral steroids for 3 years. X-rays showed deformed arthritic scaphoid, lunate and capitate. A proximal row carpectomy was performed to provide functional wrist mobility to the patient. Histopathological examination was consistent with Gorham's disease. In a patient with systemic disease receiving steroid therapy Gorham's disease poses a diagnostic challenge.

1. Introduction

Osteolysis of bone is a localized resorption, usually associated with a secondary cause like sudecks dystrophy, gout, parathyroidism, diabetes mellitus, tumours, rheumatoid arthritis, psoriatic arthritis, prolonged steroid therapy, bony aneurysm and cystic angiomas of bone.¹ The Gorham Stout syndrome is a rare idiopathic condition with osteolysis of a single bone or a group of contiguous bones around a focus. It commonly affects the skull, shoulder and the pelvic girdle.^{1–4} Isolated involvement of carpal bones is not reported in literature. (see Table 1)

The disease is known to occur across all age groups with no particular predilection to sex or race. There is no case reported with family history of a similar condition.⁵

The affected bone is known to disappear completely in stages until spontaneous remission of the condition occurs.^{5,6} During the acute phase, the patient presents with progressive pain, swelling, deformity, joint contractures, often preceded by a history of trauma.⁵ The etio-pathogenesis remains unclear but often ends with severe deformity and functional disability to the patient.⁵

Congenital Adrenal Hyperplasia is a genetic disorder associated with 21 alpha hydroxylase deficiency. Hormone production including cortisol and aldosterone is severely hindered causing feedback associated overstimulation of the adrenal glands and resultant hypertrophy. This in return causes pathological over-production of hormones such as androgens. Steroid therapy is an important form of treatment of this condition.

We report a case of Gorham's disease in a patient with congenital

adrenal hyperplasia on steroid therapy with isolated carpal bone involvement.

2. Case report

A 14 year old male presented with history of pain in the right wrist joint since four months. He was a known case of Congenital Adrenal Hyperplasia (CAH) receiving oral steroid therapy for three years. There was no history of any trauma anytime during or before the onset of pain.

On clinical examination tenderness was noted over the scapho-lunate region. Wrist movements were restricted. He was unable to perform his daily activities using the affected limb.

Radiological assessment showed osteolysis with deformed carpal bones involving the scaphoid, lunate and triquetrum. In view of his primary condition and steroid therapy, a preliminary diagnosis of avascular necrosis was suspected Fig. 1A,B. The radiographs of the opposite wrist were normal Fig. 2. Computed Tomography (CT) scan of the involved wrist showed similar features. Fig. 3A,B.

Patient was initially treated with a brace. Since there was no clinical improvement, he was advised a salvage procedure in view of gross destruction of the proximal carpal bones and no option for reconstruction. Patient wanted a painless wrist with some mobility. Hence a proximal row carpectomy was performed. The deformed and arthritic carpal bones were excised and sent for histo-pathological examination Fig. 4A,B Fig. 5A,B.

Histopathological examination of the specimen revealed fibro-collagenous tissue with numerous proliferating capillaries and

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<https://doi.org/10.1016/j.jor.2020.10.001>

Received 29 June 2020; Received in revised form 5 August 2020; Accepted 2 October 2020

Available online 9 October 2020

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Table 1
Review of literature of Gorham-Stout disease involving the hand.

Authors	Details
Beals and bird, ⁸ 1975	Reported one case of bilateral carpotarsal involvement. This patient had no nephropathy or any hereditary pattern of inheritance.
Tookman, Paice and White, ⁹ 1985	Reported a patient with osteolytic lesions of the metacarpal and metatarsal bones and terminal phalanges.
Hardegger et al ¹⁰ ,1985	Reported a case of massive osteolysis involving bilateral carpal and tarsal bones, progressing to bilateral elbows over a period of 6 years.
Garvanos and Lunn ¹¹ ,1996	Described a patient with multi-centric involvement of the hands (mainly phalanges) and feet, normal renal function, no genetic pre-disposition
Mrutyunjaya et al ¹² , 2015	Osteolysis of the middle finger, treated by ray amputation of the finger

Fig. 1. Radiographs of the wrist showing osteolysis with deformed scaphoid, lunate and triquetrum.

Fig. 2. Radiographs of the opposite wrist showing normal carpal bones.

lymphatics, consistent with Gorham’s disease [Fig. 6]. The post operative period was uneventful.

The patient was followed up 6 weeks, 3 months, 6 months, 1 year and 2 years. At 2 years follow up patient had full painless range of motion at the wrist joint. He was able to perform all his routine activities with no complaints Fig. 7A,B Fig. 8A,B. There was no evidence of involvement of any other site.

Fig. 3. Computed Tomography scan of the involved wrist showing similar features.

3. Discussion

This patient was a known case of congenital adrenal hypoplasia on steroid therapy. Hence a preliminary diagnosis of avascular necrosis of the scaphoid, lunate and triquetrum were made. Xrays and CT scan were suggestive of the same. However the histopathological examination revealed features of Gorhams disease. The differentiation between primary and secondary osteolysis could only be established by histopathological examination.

The exact pathogenesis of Gorham’s disease is still unknown. Several authors have attributed the presence of osteoclasts causing the osteolysis.^{5,6} Several others propose a theory of angiomasitosis of blood vessels and lymphatics responsible for the bone resorption. They did not find osteoclasts in the histo-pathological picture.^{7,8}

In this patient the angiomasitosis was noted in the biopsy specimen. Hence the hypothesis suggested in this report is that the Gorhams disease progresses in stages. The initial osteoclastic resorption of bone is then replaced by the angiomasitosis. This in return leads to the increased hydrolytic enzyme activity causing the osteolysis.⁷

Idiopathic osteolysis involving various sites have been described in the available literature. Beals and Bird have reported one case of bilateral carpotarsal involvement. This patient had no nephropathy or any hereditary pattern of inheritance.⁸ Tookman, Paice and White observed a patient with osteolytic lesions of the metacarpal and metatarsal bones and terminal phalanges.⁹ Garvanos et al. have described a patient with multi-centric involvement of the hands and feet. Isolated unilateral carpal bone involvement has not been described in literature. Such presentation poses a diagnostic and therapeutic challenge.

Various reports of osteolysis with multicentric involvement and associations with nephropathy have been mentioned in the literature.^{2–4} Hardegger et al. have classified osteolysis into five different types.¹⁰

3.1. Type

1. Hereditary multicentric osteolysis with dominant transmission
2. Hereditary multicentric osteolysis with recessive transmission
3. Non-hereditary multicentric osteolysis with nephropathy
4. Gorham’s massive osteolysis
5. The Winchester syndrome

This report fits into the Gorhams massive osteolysis syndrome. Our patient has monocentric involvement with no family history of the same or associated nephropathy. At follow ups up to 2 years no other site

Fig. 4. Through a dorsal approach to the wrist a proximal row carpectomy was performed and the deformed bones sent for histo-pathological examination.

Fig. 5. Post-operative radiographs of the wrist.

Fig. 7. Two years follow up x rays showing no evidence of recurrence.

Fig. 6. Histopathological examination of the scaphoid, lunate and triquetrum showing evidence of Gorham-Stout disease.

was known to be involved.

The treatment modalities for Gorham's disease are based on the site of involvement. Excision of the involved bone when feasible, radiotherapy, endoprosthetic replacement and even amputation have been described in progressive cases. In this patient with regards to the age and functional status of the patient, the pathological bones were excised and a proximal row carpectomy was performed. The patient had painless functional range of motion at 2 year follow up.

4. Conclusion

- In a patient with systemic disease receiving steroid therapy Gorham's disease closely co-incides with avascular necrosis of bone and hence poses a diagnostic challenge.
- Histopathological examination of the biopsy specimen holds key in confirmation.

Fig. 8. Full painless ROM at wrist at two years follow up.

- Excision of the pathological bone focus whenever feasible shall hold key in treatment and functional integration.

Declaration of competing interest

No conflict of interest was noted between any of the authors.

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