

Orthotic Management with a Customized Humeral Brace for Gorham-Stout Disease of the Humerus: A Case Report*

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ABSTRACT

Gorham-Stout disease is characterized by massive osteolysis or “vanishing bone” on radiograph. Due to its rarity, no standard Physical Medicine and Rehabilitation (PM&R) management has been published. With this comes the dilemma of managing another case of vanishing right humerus in a 13 year-old male, right handed student, with normal growth and development. To date, this could be the third documented case in the Philippines, but the first with humeral involvement, and the first to manage using a customized humeral brace. The absence of the right humerus affects the bimanual overhead and tabletop activities of the patient, for which a custom-made humeral orthosis was provided to manage the limited activities. There were improvements in activities such as writing, card turning, stacking, and lifting objects of variable weights, as well as with hand dexterity as evidenced by the standardized hand function tests done prior and post brace fitting. Being a rare bone disease with no standard management and unpredictable course, cases are managed symptomatically. For this case of an absent humerus significantly affecting upper extremity function, orthotic management is one aspect that could be recommended to achieve positive functional outcomes.

Keywords: Gorham-stout disease, massive osteolysis, disappearing bone disease, idiopathic multicentric osteolysis, orthotics, braces

INTRODUCTION

Gorham-Stout disease (GSD), also known as vanishing bone disease as well as idiopathic massive osteolysis, is a rare disease with its cause remaining to be obscure, but is most often characterized by endothelial proliferation of lymphatics and vessels or progressive dissolution of contiguous osseous structures where no

regeneration is noted following the osteolysis.¹ Apart from its etiology being unknown, its course has also been reported to be unpredictable based on the limited studies available. Most reports have shown that the usual presentation starts with pain, subsequent swelling, and gradual functional impairment of the affected region. One reported case was diagnosed after a pathologic fracture.^{2,3}

The prognosis of patients with GSD, like its course, is also unpredictable and diverse. The following factors affect its prognosis: extent of the bone resorption, the specific involved part and their related structures. With regards to management, there is presently, no consensus in the medical literature on what is the most effective treatment, and more often, the treatment is directed towards specific symptoms.⁴

We report a first case of idiopathic massive osteolysis of the humerus in the country as well as the first reported case of orthotic management using a customized humeral brace with the aim of improving upper extremity function and activities of daily living.

CASE

Patient Information

A 13-year old male, left handed student, was previously diagnosed with probable idiopathic massive osteolysis of the humerus, otherwise known as Gorham-Stout disease on the basis of a history of bone pain, swelling, and limitation of range of motion of the right arm with no precedent infection and a radiograph showing absence of the right humerus. The child was well until he was diagnosed with humeral fracture following an unreported fall at age 5 in 2010. Consultation was done when his hand started to swell and progressively affecting the right shoulder. On follow

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up, there was no note of callus formation on radiograph of the right humerus. In the interim since initial consult, radiographs of the right humerus revealed progression of osteolysis and no signs of bone healing. He could no longer move his right shoulder fully which primarily became the point of concern for the patient and the family. There was no note of any family history of bone disease or cancer.

Clinical Findings

Over time, there was noted decreasing muscle bulk of the deltoid, trapezius, and triceps and further reduction of right shoulder range of motion and documented resorption of the right humerus, affecting the patient's activities that required overhead reaching.

Timeline



Diagnostic Assessment

Work ups revealed no signs of infection, nor endocrine and renal dysfunctions. He was lost to follow up after the diagnosis of idiopathic massive osteolysis of the right humerus was made.

Serial radiographs showed continuous resorption of the right humerus (Figure1).

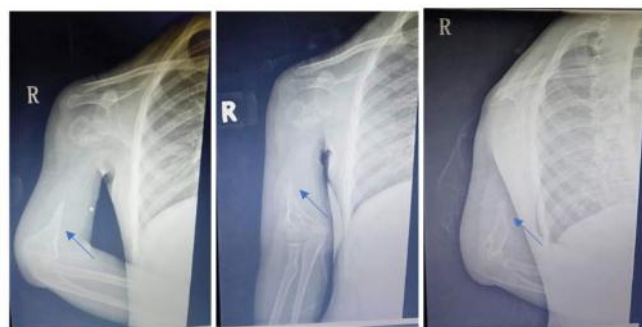


Figure 1a "Rat-tail" appearance of the remaining humerus (1a Antero-posterior view with right elbow flexed, 1b with elbow in neutral position, 1c with elbow flexed and adducted)

Therapeutic Intervention

The limitations in doing overhead activities with his right shoulder prompted consultation for possible orthotic management. He was seen by both orthopedic surgeon and pediatrician. Both specialists did not consider surgical management, radiotherapy nor anti-resorptive medications.

The patient adapted to his impaired right humeral function with the use of one hand technique in carrying simple tasks although requiring proper positioning of the wrist. He was subsequently referred to study hospital in December of 2019 for possible fabrication of a brace. On initial consult, the following findings were noted: reduced muscle bulk on the right deltoid and right trapezius and upper limb length discrepancy, the left being longer by 12 cm (right = 65 cm and left = 77 cm). The patient was prescribed with a modified humeral brace with the following components: rigid polyethylene shell covering the whole arm with extension to the elbow with 45 degree angulation, with closed-cell foam lining, three Velcro straps, and three point harness (Figure 2). The standard humeral brace was modified to extend the upper trim line to anchor at the level of the clavicle to limit the motion at the shoulder to prevent traction injury. Since the patient was able to flex and extend his elbow as long as the elbow is supported, another modification was to extend the distal trim line up to the elbow to keep the elbow flexed at 45 degrees.

Follow-up and Outcomes

Objective functional tests including Jebsen Taylor Test of Hand function and Minnesota Manual Dexterity Test were used to assess his level of hand functioning.



Figure 2a (anterior) Figure 2b (posterior) Figure 2c (lateral)

The functional tests were done pre- and post-fabrication of the brace with noted improvements in terms of efficiency of performance, specifically in writing, card turning, lifting small objects, feeding, stacking, lifting objects of varying weights, with some observations of use of compensatory strategies such as shoulder elevation and tip-toeing, particularly in reaching objects past arm's length due to absence of elbow extension. The scores for each test are presented in Appendix A and Appendix B.

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There were no adverse events reported on the use of the orthosis.

DISCUSSION

Gorham's disease is a rare disorder that may affect any part of the bone, but it most commonly involves the skull, rib, pelvic girdle, and shoulder.⁵ As of recent literature, there is no standardized treatment and results are unpredictable as is the prognosis. In one study, an 18 year old male diagnosed with the disease manifesting on his humerus, was managed with an autogenous vascularized fibular graft with wide excision and followed up after 10 years, and showed evidence of bone union and no signs of osteolysis which could be another avenue worth pursuing with research.⁶ With this case where no surgical intervention was done or contemplated by the patient's pediatric orthopedic surgeon, non-surgical and non-radical management was sought as compared with the few existing literature on Gorham-stout disease. The primary goal of the patient and the family was to have improvement in function using non-surgical means, of which, the modified humerus brace was able to provide in terms of improvement in performing activities of daily living that require overhead activities such as grooming, bathing, reaching, lifting objects, writing on the board, as well as in some active play and games. To the best of knowledge, there has not been any documented report on the role of an orthosis in managing the specific case.

Given the unpredictable nature of the disease, depending on the affected bone, its attachments and its functions, there could be a need to support, align, prevent, or correct deformities and improve function of the moveable parts of the body in which, orthotic management would address. It is recommended that upon identification of the bony structure that has undergone massive osteolysis, to consider whether there is a need to provide an external structural support to prevent further deficit or injury.

To conclude, Gorham's disease is a very rare bone disease that has proven to be a challenge not only in diagnosis, but also with its management and prediction of its course and most often monitored from initial suspicion and throughout their lifetime. While there is still a need to explore the reported management in literature which include surgery, bone grafts, radiotherapy, and bone resorption inhibitors such as bisphosphonates, the treatment goals of the above mentioned approaches would still be symptomatic as well as to aid in preventing further progression of the osteolysis. Other problems that need to be addressed are the personal and psychosocial factors especially since the patient was fully functioning prior to the injury. Specifically, body image issues and how the impairment affects present and future status in current home, school and or future employment status of the patient, and how the patient and his family can accept the unpredictability of their disease. Apart from the mentioned medical and surgical treatments, early rehabilitation can be offered in terms of physical therapy, occupational therapy and psychology counseling to address the deficits, modifications at home, school, work environment, psychosocial intervention, and in this case, orthotic management. For this case, orthotic management, a safe, non-invasive, supportive management, has alleviated the patient's impairments, and thus, improvement of his performance of activities of daily living requiring overhead arm functions. Therefore for other similar cases, an appropriate orthosis is an option to correct or reverse functional deficits to achieve positive functional outcomes. Regular monitoring and anticipatory management of a pediatric patient using patient-centered multi-disciplinary team approach is warranted.

INFORMED CONSENT

Informed consent from the parent and assent from the patient were obtained with purpose, anonymity, risks and benefits explained.

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Appendix A. Jebsen Taylor Test of Hand Function with and without Humeral Brace

Jebsen Taylor Test of Hand Function				
	With sling	Without sling or custom-made humeral brace	With custom-made humeral brace	Normative values
Writing	42.79 (manuscript) 56.49 (cursive)	39.09s (in cursive)	35.53s (in cursive)	12.2±3.5
Card turning	12.32s	6.92s	6.32s	4.0±0.9
Lifting small objects	12.5s	9.43s	12.50s	5.9±1
Stacking checkers	7.15s	7.53s	5.98s	6.4±0.9
Lifting large heavyweight objects	9.20s	9.27s	8.23s	3.3±0.7
Lifting large lightweight objects	6.45s	7.18s	6.35s	3.0±0.4
feeding	12.52s	27.39s	23.27s	3.0±0.5

*s-seconds

Jebsen Taylor Test of Hand Function

The test assesses a broad range of uni-manual hand functions required for activities of daily living divided into 7 subtests performed on both non-dominant and dominant hands with the maximum time allotted per subtests being 120 seconds. The lower the score, the greater the function, and speed, not quality of performance is measured.

Appendix B. Minnesota Manual Dexterity Test with and without Humeral Brace

Minnesota Manual Dexterity Test			
	Without sling or custom-made humeral brace	With custom-made humeral brace	Normative range
Placing	105.84s	119.94s	100-200s
Turning	56.115s	54.995s	60-90s

The Minnesota Manual dexterity test is used to measure a subject's simple but rapid eye hand coordination as well as arm-hand dexterity. In general, it measures gross motor skills. The faster the patient is able to complete a task, the better the dexterity.