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Tophaceous Gout in a Patient with Beta Thalassemia: A Case Report

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Abstract **Case Report**

Thalassemia refers to a group of chronic microcytic hereditary hemolytic anemias characterized by a defect in hemoglobin synthesis. Beta-thalassemia is more prevalent among individuals of Mediterranean, Middle Eastern, South Asian, or Indian descent. Symptoms arise from anemia, hemolysis, splenomegaly, medullary hyperplasia, and, in cases of multiple transfusions, iron overload. Diagnosis relies on genetic and quantitative hemoglobin analysis. This article presents an interesting case with images of chronic tophaceous gout in a patient with beta-thalassemia.

Keywords: gout, thalassemia, beta-thalassemia, association, tophus, heredity, anemia.

INTRODUCTION

Thalassemia is a hereditary disease characterized by chronic hemolysis and ineffective erythropoiesis resulting from defects in the globin component of red blood cell hemoglobin [1]. Intermediate and major beta-thalassemias are characterized by reduced production or total absence of beta-globin chains that make up hemoglobin [2].

On the other hand, gout is the most common inflammatory rheumatism worldwide, especially in elderly men. It results from chronic hyperuricemia with joint deposits of uric acid crystals. Additionally, a tophus corresponds to a tissue deposit of uric acid crystals surrounded by inflammatory cells during the chronic phase of the disease. It is located around joints affected by gout, preferentially in the ear (pinna and helix), olecranon bursa, Achilles and patellar tendons, distal interphalangeal joint, or digital pulp.

An increase in uric acid production in thalassemia was observed in the first case reported in 1970 by Paik *et al*., [3]. We present images of a case of chronic tophaceous gout in a patient with beta-thalassemia.

THE CASE REPORT

This is a 67-year-old man, followed in hematology for 10 years for beta-thalassemia, previously treated with erythropoietin (erythropoietin beta 5000 IU) for 8 years before its discontinuation 2 years ago. He has a family history of similar cases in a brother and two sisters, resulting from consanguineous marriage. For the past 30 years, he has been experiencing pulsatile pain in the right big toe and mechanical knee pain. He consulted several general practitioners who treated him with analgesics.

In 2010, the patient developed a painful swelling in the second left finger, leading him to consult a traumatologist. The diagnosis of tophaceous gout was made due to hyperuricemia at 136, associated with the presence of a tophus at the second proximal interphalangeal joint. He was treated with colchicine and Zyloric 300 mg.

The course of the disease has been marked by a progressive exacerbation of clinical symptoms, with extension of tophi due to poor therapeutic adherence and non-compliance with the hypouricemic diet. He then presented to the rheumatology department with chronic inflammatory polyarthralgia, joint swelling, and no axial involvement.

The clinical examination revealed a peripheral joint syndrome with a joint index of 24 involving both ankles and a synovial index of 5. Multiple tophi were observed in the metacarpophalangeal, metatarsophalangeal, proximal interphalangeal, and distal interphalangeal joints, as well as in both elbows. Additionally, there was generalized cutaneous-mucosal pallor and splenomegaly on abdominal examination.

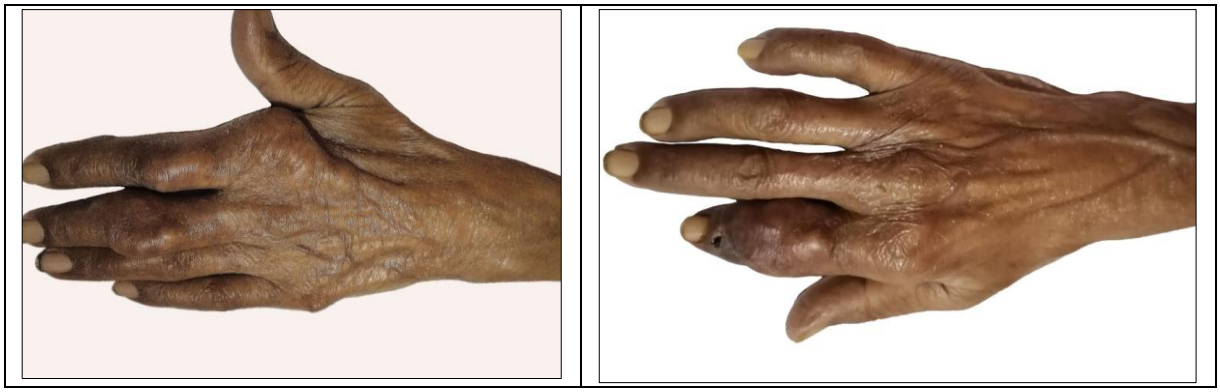

Figure 1: Multiple tophi observed in the metacarpophalangeal, proximal interphalangeal, and distal interphalangeal joints.

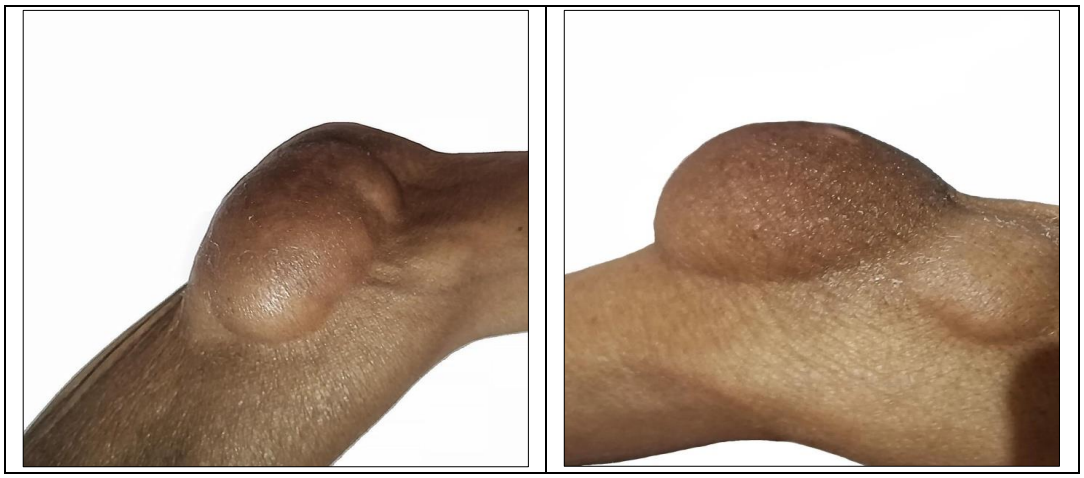

Figure 2: Tophus observed at the level of both elbows.

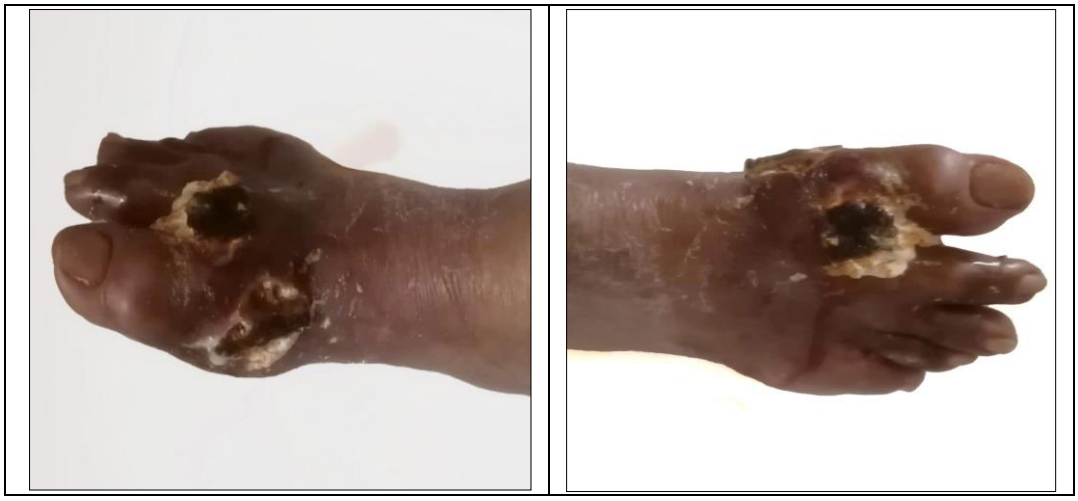

Figure 3: Appearance suggestive of an infected tophus at the level of the first metatarsophalangeal joint.

In the laboratory results, a hypochromic microcytic anemia was observed, with a hemoglobin level of 5.3. The white blood cell count was 4550, platelets were 236,000, and uric acid was 105.

X-rays of the hands and feet were performed, revealing thickening of the soft tissues with dense nodules around the joints, corresponding to tophi. There were also signs of a "barbed-wire" appearance, overall joint narrowing, diffuse demineralization, and multiple erosions and geodes.

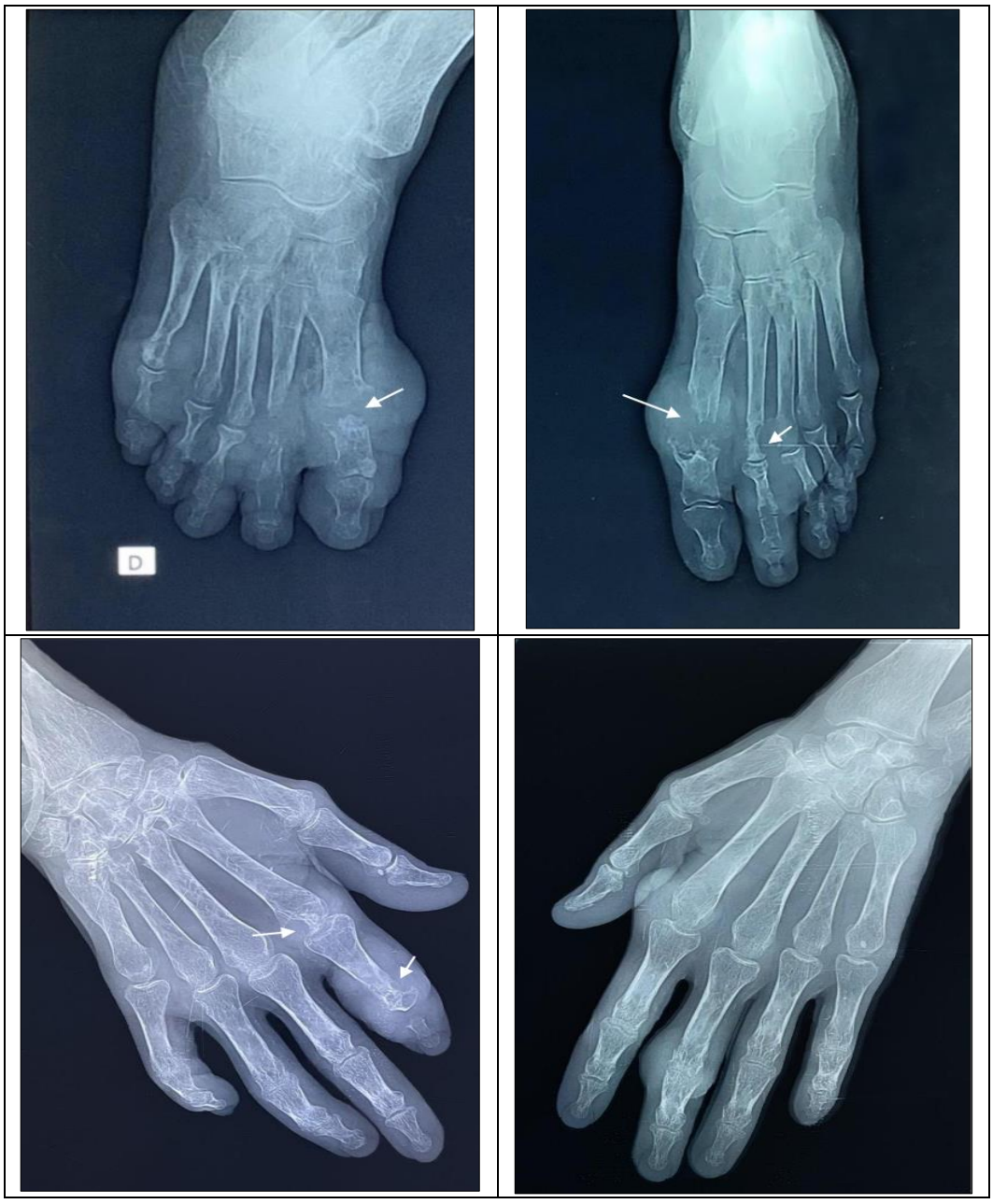

Figure 4: Standard X-rays of both forefeet and both hands, frontal view, showing typical signs of gout.

The patient was treated with colchicine 1 mg, Zyloric 700 mg, and received a transfusion of two units of red blood cells, in addition to a hypouricemic diet. After 3 weeks of treatment, there was clinical improvement with reduced pain and regression of synovitis. On the follow-up assessment, the uric acid level was 55.61, indicating improvement compared to previous results.

DISCUSSION

Beta-thalassemia (or beta thalassemia) is a genetic disorder of hemoglobin, a substance found in the red blood cells that transports oxygen throughout the body. Beta-thalassemias vary in severity: some forms are asymptomatic, while others can be life-threatening.

This condition is characterized by a reduction in the synthesis of the beta chain and inadequate formation of HbA. However, the synthesis of the alpha chain continues without alteration, forming unstable aggregates in red blood cells and causing inadequate and inefficient erythropoiesis. These red blood cells are destroyed in the bone marrow or spleen. This increased destruction of immature erythrocytes leads to an

increased production of uric acid, a final product of purine metabolism. Uric acid tends to crystallize in various tissues when serum is supersaturated, at around 7.0 mg/dl, and beyond this level, the risk of gout or kidney stones is increased.

A recent cross-sectional study conducted in Thailand showed that the prevalence of hyperuricemia in the thalassemia cohort was approximately 40%, but gouty arthritis accounted for only 6% of the study population. Studies suggest that the relatively high rate of red blood cell turnover in thalassemia may be related to a higher level of endogenous uric acid or serum uric acid.

However, elevated serum levels of uric acid in patients with thalassemia can sometimes lead to hyperuricosuria, causing microscopic hematuria, renal tubular dysfunction, and abnormalities in glomerular filtration rate (GFR). This renal dysfunction with prolonged hyperuricemia eventually leads to the development of gouty arthritis. Kumar *et al*., confirmed the first case of crystal-proven gout in thalassemia patients and concluded that long-term viability and renal failure had inevitably favored the development of gout in this patient.

However, the presence of tophaceous gout is distinctly unusual, with only a few isolated case reports available so far.

CONCLUSION

Hyperuricemia is often observed in patients with thalassemia due to increased cellular turnover, but a low percentage of these patients develop gout, which can occur due to renal dysfunction. Further detailed studies are needed to better understand the underlying biological mechanisms of this association.

Consent: Verbal informed consent was obtained from the patient for their anonymized information to be published in this article.

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