Tophaceous Gout in a Patient with Thalassaemia Presenting as Multiple Soft-tissue Masses

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Abstract

Although patients with haemolytic haemoglobinopathies characteristically are over-producers of urate, and hyperuricaemia is frequently recognised, tophaceous gout has rarely been reported in such patients.

We report a 40-year-old man with thalassaemia who developed nodules on hands and feet with a history of arthropathy. In laboratory investigation, blood chemistry showed marked hyperuricaemia. Excisional biopsy confirmed the diagnosis of tophi.

Gout deposits in skin (tophi) are uncommon. The aim of this report is to emphasise the importance of considering this disease entity in the differential diagnosis of soft-tissue lesions in a thalassaemic patients with chronic arthritis.

Key words: Gout, tophi, thalassaemia.

Introduction

Gout is the common end-point of a group of disorders that produce hyperuricaemia. It is marked by transient attacks of acute arthritis to chronic gouty arthritis and the deposition of masses of urates in joints and other site, creating tophi. The various conditions producing hyperuricaemia and gout are divided into primary and secondary gout. Primary gout in which the basic metabolic defect is unknown consist of 90% of cases. Secondary gout in which the cause of hyperuricaemia is known, e.g., intrinsic renal disease, drugs, starvation, myeloproliferative disease, haemolytic anaemia, etc., is less common (10% of cases). Secondary tophaceous gout in haemolytic haemoglobinopathies presenting as multiple soft-tissue masses is unusual. By reviewing the medical literature, only few such cases have been reported.

We report a 40-year-old male with thalassaemia intermedia, who presented to our hospital with tophaceous gout and chronic arthritis to emphasise the importance of considering this disease entity in the differential diagnosis of a soft-tissue lesion.

Case history

A 40-year-old man was referred for evaluation of painless multiple masses on dorsal and palmar aspect of hands and dorsal aspect of feet. The patient had history of thalassaemia. By reviewing his previous medical records, the patient was found to be a case of thalassaemia intermedia diagnosed in childhood. There was a history of multiple blood transfusions since childhood. His mother and sister had died of thalassaemia. He had experienced recurrent swelling, severe pain of joints of feet and hands occurring three or four times a year since 15 years, and had been treated symptomatically. Acute attacks of arthritis were self-limiting and resolved within two to three weeks. He denied any symptoms of trauma and had no history of renal disease.

Physical examination revealed anaemia and

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splenomegaly. Also the patient developed multiple masses two to three cm in diameter on the dorsal and palmar aspect of hands, and dorsal aspect of feet. The masses were freely mobile and did not appear attached to the underlying bony skeleton. At the time of examination, the joints were normal. Laboratory studies showed normal urinanalysis, serum electrolytes, iron profile, and absence of rheumatoid factor and antinuclear antibodies. His uric acid level and ESR were high (uric acid was 9.6 mg/dl and ESR was 45 mm/h). He had haemoglobin of 5.4 gm%. His blood film showed microcytic hypochromic red cell morphology, marked anisopoikilocytosis, basophilic stippling, presence of target cells, tear drop cells, and normoblasts. His blood urea level and serum creatinine were within normal limits. Plain film radiograph revealed decreased bone density and mild erosive changes at the tarsal, metatarsal, metacarpal, and phalangeal bones. An excisional biopsy was performed. The resected specimen was well circumscribed involving the dermis and subcutaneous tissue. Histopathologically, skin biopsy showed foci of brown crystalloid deposits. Also, there were some areas of amorphous, pale staining deposits surrounded by mononuclear cells and multinucleated giant cells. The overlying epidermis was hyperplastic and showed parakeratosis and hyperkeratosis on its surface. On polaroscopic examination, the needle shaped crystals revealed negative birefringence.

One unit of blood transfusion was given to treat anaemia. The patient was prescribed allopurinol 300 mg daily, folic acid supplement, and colchicine prophylaxis in doses of 0.6 mg one time daily. The patient was discharged 12 days after admission in a stable condition.

**Discussion**

Monosodium urate gout is a metabolic disease. It is typically associated with an increased uric acid pool, hyperuricaemia, episodic acute and chronic arthritis, and deposition of MSU crystals in connective tissue tophi and kidneys.

Secondary gout in haemoglobinopathies is uncommon. Although patients with haemolytic haemoglobinopathies characteristically are over-producers of urate, and hyperuricaemia is frequently recognised, clinical gout has rarely been reported in such patients. With increasing lifespan of patients with thalassaemia and likelihood of increased occurrence of renal function...
abnormalities, it is anticipated that gout will more frequently be responsible for joint symptoms and tophi formation in such patients. But our patient had no renal function abnormalities.

Tophi are formed by aggregation of urate crystals. Tophi may appear in the articular cartilages of joints and also in the periarticular ligaments, tendons, and soft tissues. Less frequently, they may appear in the kidney, nasal cartilages, skin of the fingertips, palms or soles, as well as elsewhere. Superficial tophi can lead to ulceration of the overlying skin as in our case too the patient developed an ulcer on the lateral aspect of the foot.

Tophi develop an average of 12 years after the initial attack of gout and present as soft tissue lumps, ulcerative skin lesions, nerve compression and/or joint destruction leading to functional and cosmetic deformity. Differentiation of tophaceous gout from malignancy can be difficult. Both can present with a mass with or without pain, night pain, or neurologic compromise from nerve involvement or compression. Plain radiographs are usually not helpful in making a diagnosis. Classic radiologic findings of gout, such as para-articular erosions, cannot be distinguished from tumour infiltration with certainty.

Gout classically causes acute, episodic, monoarticular arthritis of the first metatarsophalangeal joint. Upper limb involvement is more unusual but has been described, especially when there is extensive involvement elsewhere in the body or a long history of gout. Gout is known to mimic conditions as diverse as joint and soft tissue infections, skin malignancies, nerve compression syndromes, and soft-tissue tumours. In view of these differential diagnoses, excisional biopsy of a skin nodule was performed in our case.

The diagnosis of a tophaceous nodule is established by demonstration of monosodium urate crystals by fine needle aspiration cytology or biopsy. The definitive diagnosis can be made by identification of the characteristic negative birefringent needle-shaped crystals obtained from tophi.

In summary, when the clinical history and the presence of hyperuricaemia suggest tophaceous gout, the definitive diagnosis can be made by identification of the characteristic negative birefringent needle-shaped crystals. We have here described a case of an uncommon presentation of a common disease due to the uncommon cause to emphasise the importance of considering this disease entity in the differential diagnosis in a patient with chronic arthritis and soft-tissue lesion.

References