Chronic Tophaceous Gout in a Patient with Cyanotic Congenital Heart Disease

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Abstract

Introduction: Gout is a clinical syndrome resulting from deposition of urate crystals in joints causing inflammation, intense pain and even disability. In 2003 National Nutrition and Health Survey (NNHeS), the overall prevalence of gout was 1.6% and the prevalence of hyperuricemia was 28.4% of the 4498 adults >20 years old. Gout complicating cyanotic congenital heart disease (CCHD) was infrequently recognized in early reports with Somerville referring only nine patients over a period of five years in the registry of two hospitals. Recent studies have shown increased recognition, with Yamamura identifying 16% of the 92 patients with CCHD to have gouty arthritis. There is no local data on the frequency of gout among patients with CCHD

Case Presentation: 26-year-old male previously diagnosed with Transposition of Great Arteries presented with a mass on the 5th digit of left hand with pathologic bone destruction on radiograph underwent disarticulation of the same finger. Intraoperative findings revealed chalky substances that led to the referral to rheumatology section. A diagnosis of chronic tophaceous gout was considered after review of history. He had a three year history of intermittent polyarticular arthritis involving the elbows, knees and ankle joints and later developed tophi in the helices of the ears and the DIP joint of the fifth digit of the left hand. There is increasing level of hyperuricemia as the severity of arthritis progressed. He had a polyarticular gout flare post operatively. The gout attack resolved after treatment with colchicine. Allopurinol was initiated after control of gout flare with resulting decrease in uric acid level.

Discussion: This case described a young male patient with transposition of great vessels with acute intermittent polyarthritis and tophi formation. The occurrence of gout and the complexity of the congenital heart disease have caused a considerable financial, functional and emotional instability on the patient, his parents and extended family. In patients with cyanotic congenital heart disease, the prevalence of hyperuricemia ranges from 16 to 43%. Several factors identified to be associated with hyperuricemia in CCHD in our case which include hypoxemia, polycythemia and impaired renal function. Nonetheless, management of gout in patient with CCHD is similar to the primary and secondary gout.

Conclusion: Gouty arthritis should be a differential in a patient with a cyanotic congenital heart disease complaining of joint swelling and tenderness. Early and prompt diagnosis will relieve symptoms and prevent complications that may lead to physical inactivity of patients.

Keywords: gout, congenital heart disease, arthritis, case report

Introduction

Gout is a clinical syndrome resulting from deposition of urate crystals in joints causing inflammation, intense pain and even disability in patients. The data on the prevalence of gout locally and in developing countries show increasing prevalence. In 2003 National Nutrition and Health Survey (NNHeS), the overall prevalence of gout was 1.6%[1] and the prevalence of hyperuricemia was 28.4%[2] of the 4498 adults >20 years old. Over a 10-year period observational study of adults, the overall prevalence of gout increased from 2.9 per 1000 in 1990 to 5.2 per 1000 in 1999.[3] In a more recent Nutrition and Health Survey in Taiwan (NAHSIT), the prevalence of gout has increased from 4.47% to 8.21% in men and 2.19% to 2.33% in women from a 1993-1996 survey to a 2005-2008 survey.[4] Likewise, the US National Health and Nutrition Examination Survey 2007-2008 showed increasing prevalence of gout to 3.9% compared to NHANES III (1988-1994) which showed a previous of 2.7%.[5] In a systematic review and meta-analysis on prevalence of gout and hyperuricemia in China showed pooled prevalence of hyperuricemia of 13.2% (95% CI; 11.9%,14.6%) and the pooled prevalence of gout of 1.1% (95% CI; 0.7%, 1.5%).[6]

Gout complicating cyanotic congenital heart disease was infrequently recognized in early reports with Somerville referring only nine patients over a period of five years in the registry of two hospitals.[7] Recent studies have shown

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increased recognition, with Yamamura[8] identifying 16% of the 92 patients with cyanotic congenital heart disease to have gouty arthritis. Among these cyanotic heart diseases, congenitally corrected transposition of the great arteries (ccTGA) account to one percent.[9] With this, gout is rarely noted as a clinical problem. This paper aims to report a first case of chronic tophaceous gout in flare in a patient with congenital heart disease recognized only after disarticulation of the 5th digit of left hand.

Case Presentation

A 26-year-old man with cyanotic heart disease was referred due to an intraoperative finding of a chalky substance on the disarticulated fifth digit of left hand. (Figure 1) The patient was diagnosed transposition of great arteries at birth. He has undergone several cardiac operations including Balloon Atrial septostomy, at ages 10 & 11 years old and subsequent Blalock Taussig Shunt at age 12 years old. He was compliant with his cardiology consultations and has undergone several phlebotomies due to symptomatic polycythemia.

However, three years prior to this referral, he developed acute intermittent polyarticular asymmetric arthritis involving

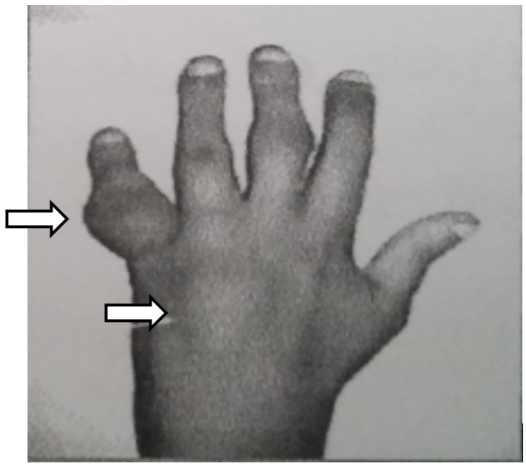

Figure 1. Tophi on 5th digit and dorsum of left hand pre operative

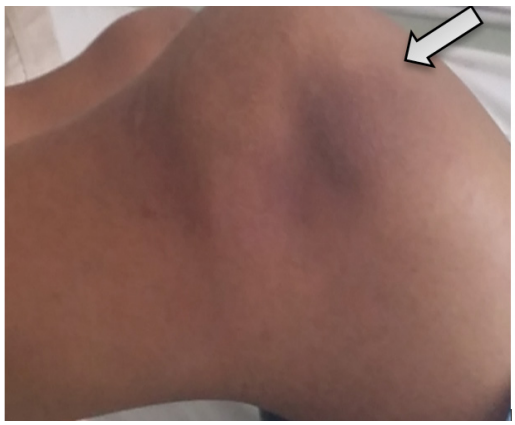

Figure 2. Swollen(arrow),warm and tender left knee

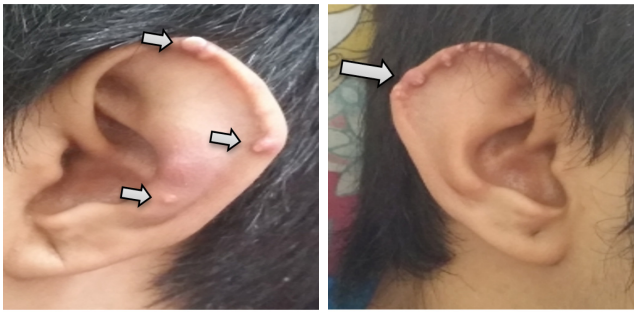

Figure 3. Multiple Tophi on the pinna of left and right ear, respectively

the elbows, knees (Figure 2) and ankles which he self-medicated with paracetamol and tramadol with some relief. The arthritis led to limitation of physical activity. Blood Uric acid at that time was 0.40mmol/L. Approximately a year later, he developed tophi on the DIP of 5th digit of left hand.

Four months prior to this admission, he had pain of the 5th digit of left hand, the xray showed a probable bone tumor with pathologic fracture. He underwent disarticulation of this finger. Pre-operative preparation included hematology evaluation due to the secondary erythrocytosis and cardiology evaluation for the congenital heart disease. Pre-operative laboratories showed erythocytosis (hgb:190 g/L and hct:0.60) hyperuricemia (blood uric acid 0.78mmol/L); urinalysis showed acidic urine with proteinuria, microscopic hematuria and granular casts. Serum creatinine was 118 umol/L with eGFR of 73ml/min/1.73m2. Ultrasound of left hand showed a fairly defined, isoechoic to subcutaneous tissue focus confined to the subcutaneous layer measuring approximately 2.5x 3.2x3.4 cm which was not continuous with the calcific mass.

He was referred to the rheumatology section due to an intraoperative finding of chalky substance in the tendon sheath and on the mass excised on the 5th digit of left hand. On physical examination, he has multiple tender and swollen joints with multiple minute tophi in the helices of both ears. (Figure 3) There is clubbing of fingers and toes. Repeat blood uric acid was 0.86 mmol/L. Biopsy report revealed that the excised mass was consistent with gouty tophus. He was treated with colchicine 500mcg/tab once a day with improvement of the gout flare. Allopurinol 100mg one tablet once a day was initiated on resolution of gout flare. Subsequent follow ups showed decline on blood uric acid level with no recurrence of gout flare.

Discussion

This case described a young male patient with transposition of great vessels with acute intermittent polyarthritis and tophi formation. His arthritis was not diagnosed to be gout until he underwent disarticulation of the fifth digit of left hand. Intraoperative findings revealed chalky substance that led to the referral to rheumatology

section and a diagnosis of chronic tophaceous gout was considered after the review of history.

The clinical presentations are compatible to gout based on the ACR criteria and are similar to those of primary and secondary gout. The appearance of tophi on the helices of both ears and on the disarticulated 5th digit of left hand is not an uncommon presentation.[12]

The occurrence of gout and the complexity of the congenital heart disease have caused a considerable financial, functional and emotional instability on the patient, his parents and extended family. Several studies[10,11] stated that the higher level of blood uric acid and greater attack frequency will lead to higher medicine cost and probable hospital admission; likewise the occurrence of tophi and co morbid conditions impair functional and health related quality of life. Parents of 20 patients with congenital heart disease have described an increased out-of-pocket costs and absences from work during hospital stay.[14] The pathology of gout is the deposition of urate crystals in joints from long standing hyperuricemia.[13] In patients with cyanotic congenital heart disease, the prevalence of hyperuricemia ranges from 16%[15,17] to 43%.[16]

Several factors identified to be associated with hyperuricemia in Cyanotic Congenital Heart Disease (CCHD) in our case which include hypoxemia, polycythemia and impaired renal function.[8,15,16] Despite undergoing Blalock Taussig Shunt, there was a considerable cyanosis in this patient with the evidence of clubbing of fingers and polycythemia in routine laboratories. Chronic hypoxemia in CCHD was found to be a strong predictor of GFR[20] and causes secondary erythrocytosis[9] which will further cause blood hyperviscosity. The hyperviscosity in turn increase the efferent glomerular arteriolar resistance, hydraulic pressure across the glomerulus and filtration fraction which would result in an increase in oncotic pressure in the post glomerular vessels promoting fluid and solute reabsorption and fluid retention.[19] This patient's hemoglobin level was almost always >130mg/dL and Hematocrit >40 for which he has undergone several phlebotomies. The occurrence of hyperuricemia came later in his life probably due to compensatory polycythemia.

In a study of 10 patients with cyanotic heart disease by Ross et al.[18], high plasma uric acid levels were secondary to inappropriately low fractional uric acid excretion, not to urate overproduction. Supported by necropsy study of 13 patients with cyanotic congenital heart disease without other extracardiac co morbidities[20], vascular and non-vascular glomerular abnormalities were noted. In a population of patients with CCHD, GFR was <90ml.min.1.73m2 in 65.7%.[17] The presence of proteinuria in our case, however,implies intrinsic renal damage and is a typical manifestation of glomerular sclerosis.

In this patient there is both urate overproduction secondary to increased red blood cell turnover secondary to polycythemia and intrinsic renal dysfunction leading to decreased urate excretion. As a consequence of the chronic inability to eliminate urate as rapidly as it is produced, the formation of tophi ensued. The patient had tophi formation within three years from gout onset which is not different from primary gout and other secondary gout.[12] Similar to the study of Ritchelle[13], tophaceous gout develops within five years of onset of gout in 30% of untreated patients.

The management of gout in patient with CCHD is similar to the primary and secondary gout.[12] Treatment with colchicine has dramatically relieved pain in the patient. Colchicine is an antimitotic agent decreasing the functional activity of granulocytes migrating into the inflamed area and inhibiting the release of pro-inflammatory substances hence controlling pain. The blood urate concentration is not altered with this medication hence hypouricemic agents are needed. After two weeks, upon cessation of pain, allopurinol, a xanthine oxidase inhibitor was started at 100mg one tablet OD with noted good response and decreasing uric acid level on subsequent follow up.

Conclusion

Gouty arthritis should be a differential in a patient with a cyanotic congenital heart disease complaining of joint swelling and tenderness. Early and prompt diagnosis will relieve symptoms and prevent complications that may lead to physical inactivity of patients.

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