Tophaceous gout in uncorrected cyanotic congenital heart disease

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DESCRIPTION

Gout refers to an inflammatory condition caused by deposition of urate crystals in joints leading to painful arthritis.1 Tophaceous gout occurs due to collection of urate crystals in subcutaneous and soft tissue. Unlike primary gout where the aetiology lies in malfunction of uric acid metabolism, secondary gout occurs due to excessive production of uric acid or due to abnormal handling of uric acid by kidneys.2 Tophaceous gout in cyanotic congenital heart disease (CCHD), though rare, has been well reported.

A 23-year-old man, a known case of uncorrected severe tetralogy of fallot with pulmonary atresia, presented to us with painful swellings of bilateral first metatarsophalangeal joints and interphalangeal joint of right middle finger. On examination, he was cyanosed with a resting oxygen saturation of 84% on room air and had grade IV clubbing (figure 1A). Large tophi could be seen at base of both great toes and interphalangeal joint of right middle finger, along with few tophi with spontaneous skin ulceration at ankles and planter aspects of both feet (figure 1A–D). The patient had haemoglobin of 200 g/L and uric acid was 7.8 mg/dL. His urea and creatinine were 39 mg/dL and 0.9 mg/dL, respectively, and estimated glomerular filtration rate was 111.1 mL/min/1.73 m². Other biochemical parameters including potassium (5.1 mEq/L) and lactate dehydrogenase levels (270 U/L) were within normal limits. His urine examination revealed mild proteinuria (200 mg/24 hours). He was managed with colchicine and non-steroidal anti-inflammatory drugs (NSAIDs) for acute gout and was subsequently started on allopurinol. He has been doing well on follow-up with no further flares of acute gout.

In 1961, Somerville identified secondary gout only in nine patients of CCHD over a period of 3 years,3 but today with prolonged survival of these patients, the reported incidence is much higher.4 The incidence of gout in CCHD is directly linked to degree and duration of cyanosis, resultant secondary polycythaemia and hyperuricaemia.5 The hyperuricaemia occurs not only due to secondary polycythaemia but also due to impaired kidney function. Both hyperviscosity and hyperuricaemia damage the kidneys. There may be significant glomerular sclerosis and tubular injury, with proteinuria being the only biochemical abnormality.1

Management of tophaceous gout in CCHD is similar to other types of gout.6 In acute flare anti-inflammatory drugs like NSAIDs and antimitotic agents like colchicine are used, however, in severe cases, steroids and interleukin-1 antagonist (anakinra) have been shown to be beneficial.2 After acute flare is resolved, uric acid-lowering drugs like allopurinol or febuxostat are added based on renal function. The index patient was managed as per the guidelines and there has been no recurrent flare of acute gout on follow-up.

To conclude, tophaceous gout is a rare, although well-known complication of CCHD. With improved long term survival of CCHD patients due to better healthcare facilities, we may now encounter

Patient’s perspective

I have heart disease and then developed pain and swelling on my hands and feet. After the medicine, I am good now and do not have any pain in my joints.

Learning points

► Tophaceous gout in cyanotic congenital heart disease (CCHD) is rare, though a known complication.
► The incidence of gout in CCHD is related to degree and duration of cyanosis with resultant polycythaemia, hyperuricaemia and impaired kidney function.
► Management of secondary gout in CCHD is similar to other types of gout.

Figure 1 Clubbing and tophi at interphalangeal joint of right middle finger and bilateral first metatarsophalangeal joints (A and B). Plantar tophi with ulceration of overlying skin (C). Bilateral finger and toenails clubbing with tophi (D).
secondary gout more often in these patients. The management, however, remains similar to other types of gout.

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