



Chronic tophaceous gout: a case report in PHC

Gota tofácea crônica: relato de caso na Atenção Primária à Saúde

Gota Tofácea Crônica: reporte de un caso en APS

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Abstract

Introduction: Gout is a clinical entity that humans have lived with since the 5th century BC. It stems from a metabolic change in purines, resulting in the accumulation of uric acid in the blood, with its incidence most evident between the third and fourth decades of life, predominantly in men. Risk factors include obesity, Afro-American race, male sex, and higher levels of serum uric acid, and it is also related to increased intake of red meat, seafood, fish, saturated fats and dose-dependent alcohol intake. **Case presentation:** In its evolution, it can present an exuberant case of chronic tophaceous gout in an adult patient in his fifth decade of life, without regular follow-up. In addition to discussing the effects of poorly controlled hyperuricemia, this paper portrays one of the most challenging complications in the routine of the family and community doctor, osteoarticular involvement. **Conclusions:** The case in question stands out for its relevance, given the advanced evolutionary stage, which affects the patient's quality of life and autonomy, demonstrating how practice in primary health care can be challenging and has its potential in the comprehensive approach.

Keywords: Gout; Public health; Joints.

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Resumo

Introdução: A gota é uma entidade clínica com a qual o ser humano convive desde o século V a.C. Decorre de uma alteração metabólica das purinas, resultante do acúmulo de ácido úrico sérico cuja incidência é mais evidente entre a terceira e quarta década de vida, predominante em homens. Apresenta fatores de risco como obesidade; raça afro-americana; sexo masculino; maiores níveis de ácido úrico sérico; e, ainda, relação com a alta ingestão de carnes vermelhas, frutos do mar, peixes, gorduras saturadas e consumo alcoólico dose-dependente. Em sua evolução, pode se apresentar de diferentes formas, a saber: hiperuricemia assintomática, artrite gotosa aguda, gota intercrítica e gota tofácea crônica. **Apresentação do caso:** Apresenta-se aqui um caso vultoso de gota tofácea crônica em paciente adulto, na quinta década de vida, sem acompanhamento regular. Além de discutir os efeitos da hiperuricemia mal controlada, o presente trabalho retrata uma das principais complicações mais desafiadoras da rotina do médico de família e comunidade: o acometimento osteoarticular. **Conclusões:** O caso em questão se destaca por sua relevância, haja vista o estágio evolutivo avançado que afeta a qualidade de vida e autonomia do paciente, demonstrando como a prática na atenção primária pode ser desafiadora e tem na abordagem integral sua potencialidade.

Palavras-chave: Gota; Saúde pública; Articulações.

Resumen

Introducción: La gota es una entidad clínica con la que el ser humano ha convivido desde el siglo V antes de Cristo. Se deriva de una alteración metabólica de las purinas, resultante de la acumulación de ácido úrico sérico, cuya incidencia es más evidente entre la tercera y cuarta década de la vida, predominando en hombres. Presenta factores de riesgo como obesidad, raza afroamericana, sexo masculino, mayores niveles de ácido úrico sérico y también está relacionada con la ingesta aumentada de carnes rojas, mariscos, pescado, grasas saturadas e ingesta alcohólica dosis dependiente. En su evolución, puede presentarse de diferentes formas, a saber: hiperuricemia asintomática, artritis gotosa aguda, gota intercrítica y gota tofácea crónica. **Presentación del caso:** Aquí se presenta un caso exuberante de gota tofácea crónica en un paciente adulto, en la quinta década de la vida, sin seguimiento regular. Además de discutir los efectos de la hiperuricemia mal controlada, el presente trabajo retrata una de las complicaciones más desafiantes en la rutina del médico de familia y comunidad, la afectación osteoarticular. **Conclusiones:** El caso en cuestión destaca por su relevancia, dado el estadio evolutivo avanzado, que afecta la calidad de vida y la autonomía del paciente, demostrando cómo la práctica en la atención primaria puede ser desafiante y tiene su potencial en el abordaje integral.

Palabras clave: Gota tofácea crónica; Poliarticular; Complicaciones.

INTRODUCTION

Gout is a metabolic disease that classically presents as inflammatory arthritis caused by periods of hyperuricemia and the accumulation of monosodium urate crystals in various parts of the body, such as articular cartilage, subchondral bone, synovial membrane, capsule, and areas of lower temperature, such as the superficial tissues of the extremities.^{1,2}

Gout is directly influenced by genetic and dietary factors.³ The disease mainly affects middle-aged and older men, as well as postmenopausal women, being six times more common in men.⁴ In addition, a high serum uric acid concentration is the most important risk factor for the development of gout.² However, there are cases where attacks are triggered with normal or low serum levels, although these are a minority.^{1,2}

Hyperuricemia is usually defined when serum uric acid levels are above 7 mg/dL.⁴⁻⁶ This excess of the metabolite generally originates from meals rich in meat (which increases insulin resistance, which reduces renal urate excretion), sugar, salt, or excessive alcohol consumption (which, in turn, increases urate production and reduces excretion). Problems in the elimination process, on the other hand, indicate an alteration in the kidney.^{4,5}

The disease itself is caused by urate crystals and not by urate in solution. When these crystals deposit in the joints, the tophi are recognized as antigens by the body, which then sends leukocytes to the synovial fluid. These white blood cells phagocytize the crystals, causing pain and triggering an inflammatory reaction, thus characterizing the acute phase.^{4,5}

The natural history of gout involves progression from a prolonged asymptomatic period of monosodium urate crystal accumulation in the joints, interspersed with phases of mono- or oligoarthritis, to a phase of chronic arthritis with the presence of crystal deposits, the tophi.⁶⁻⁸ This paper reports a case study conducted with a patient in the chronic gout phase.

This phase is clinically characterized by the presence of tophi, secondary to the accumulation of uric acid, protein matrix, inflammatory cells, and foreign body giant cells in tendons, ligaments, cartilage, subcutaneous tissue, and periarticular regions.^{1,2} In this scenario, imaging studies can be useful to assess the severity of the disease, the extent of depositions, and the presence of chronic inflammation. In addition, it can be a useful tool to monitor the response to uric acid-lowering therapy.^{1,7}

Regarding chronicity, patients with this condition present inflammatory mediators of both acute and chronic disease, leading to synovitis, bone erosion, and chronic synovitis.⁶⁻⁸ Therefore, a point of interest in this study is to address the extra-articular manifestations, demonstrating, in practice, possible long-term complications. That said, in the patient in question, significant consequences can already be observed, which require intervention and improved follow-up.

CASE PRESENTATION

A 56-year-old Black man presented for an outpatient visit at a primary health care (PHC) unit; his past medical history included a diagnosis of gout approximately 15 years prior and hypertension 10 years ago. The patient did not have regular medical follow-up and decided to resume care after an exacerbation of gout-related deformities, which had been limiting his autonomy and self-care. Physical examination revealed signs of arthritis and multiple joint deformities, especially in the wrists, metacarpophalangeal (MCP) joints, Achilles tendon, knees, and ankles (Figures 1–3). The lesions were painless, stony in consistency, with an irregular surface and adhered to the joints. Regarding medication, the patient was



Figure 1. Deforming arthropathy in metacarpophalangeal joints.



Figure 2. Deforming arthropathy in the metatarsophalangeal joint of the big toe (gout) and in the Achilles tendon region.



Figure 3. Deforming arthropathy in the right knee.

chronically using colchicine 0.5 mg daily and irregularly using allopurinol 100 mg. analysis revealed the following findings: uric acid 9.4 mg/dL; creatinine 2.24 mg/dL; creatinine clearance 25 mL/min/1.73 m²; and urea 96 mg/dL; among other findings without repercussions for the case. Therefore, allopurinol 200 mg daily was initiated, in addition to the other medications the patient was already using: colchicine 0.5 mg daily, losartan 100 mg daily, and amlodipine 10 mg daily. During the first evaluation, radiographs were also requested to document the aforementioned lesions using imaging methods, as well as routine laboratory tests for control and monitoring.

After approximately 45 days, the patient returned to the outpatient clinic with the test results. Laboratory tests performed on April 25, 2024, showed the following changes: uric acid 7.6 mg/dL; creatinine 3.41 mg/dL; creatinine clearance 20 mL/min/1.73 m²; and urea level 215 mg/dL. The radiographic examinations (Figures 4–6) corroborated the clinical assessment, showing periarticular porosity and erosions, mainly. The patient is being followed up by the PHC unit and has been referred for joint follow-up with a nephrologist and a rheumatologist.



Figure 4. X-ray of the right knee.



Figure 5. X-ray of both feet.

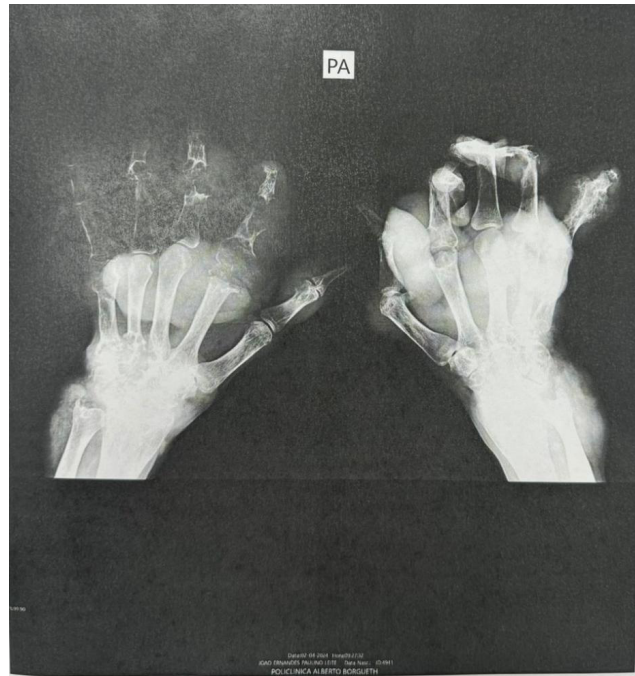


Figure 6. X-ray of both hands.

DISCUSSION

The case described involves a male patient with chronic gouty arthritis, presenting with characteristic chronic lesions that restrict and modify his daily activities, in addition to reporting renal complications due to poor medication adherence and risk factors related to daily habits.

As seen in the literature, this condition is a sensitive disease for PHC, being more prevalent in men's health care. Statistically, the overall prevalence of this disease is estimated at 1.4% of people followed in family and community medicine clinics in the United Kingdom, with a ratio of 3.6 men to 1 woman.^{1,3,9} It is expected that, in a registered population of 3,000 users, the family health team will follow approximately 7 to 9 patients with gout.¹⁰⁻¹²

Among the factors that lead to chronicity are: excessive alcohol consumption, starvation, long walks, trauma, surgical interventions, use of thiazide diuretics, blood transfusions, sudden temperature changes, and age. These factors can lead to acute crises, characterized by clinical manifestations such as pain, heat, edema accompanied by hyperthermia, chills, and immobility of joints of varying degrees. Furthermore, the recurrence of these crises leads to chronic gouty arthritis.^{2,3,6}

The clinical picture is divided into four clinical phases, the fourth being chronic gouty arthritis, which can develop articular tophi, cutaneous nodules mainly in the trabecular region, lithiasis, renal insufficiency, and cardiac and hepatic changes. The crises can be mono- or polyarticular, usually affecting the metatarsophalangeal joint of the big toe or hallux (podagra). When untreated, it can lead to painful destructive arthropathy and urolithiasis, but the latter are lesions that can be corrected through the control of hyperuricemia.^{1,4,5,8}

Histopathologically, the tophi show granulomatous reactions, being pathognomonic of the disease; they occur more frequently in digits, wrists, ears, knees, and Achilles tendon, as well as renal pyramids, heart valves, and sclera. The formation of tophi occurs because of aggregates of crystals enveloped by

a granuloma-like response, with a zone of giant cells surrounded by a fibrous layer. Erosions are located around the tophi. Urate crystals lead to effects on osteoblasts and cause reduced cell viability in these cell types, explaining bone resorption at the sites where the tophi are located.⁶⁻⁸

The gold standard for diagnosing gout is the observation of monosodium urate crystals under polarized light microscopy with compensation, where these crystals exhibit negative birefringence. The sample should preferably be collected from recently affected joints, as well as from previously affected joints. After confirming the presence of monosodium urate crystals in the joint environment, it is necessary to quantify this deposition, as well as its extent and the induced structural damage.^{4,5}

In addition to anamnesis and physical examination, imaging resources such as X-rays, ultrasound, and dual-energy computed tomography can be useful in evaluating the chronic phase of the disease and the resulting damage.^{4,5} In gout, the most characteristic radiographic findings include asymmetrical erosive arthritis, with preservation of joint space (except in late stages) and periarticular bone density.⁹

However, in the presence of a typical clinical presentation, the clinical diagnosis is reasonably accurate and acceptable in the absence of available microscopy or a rheumatologist; this diagnosis, however, is not definitive.^{4,5}

Hyperuricemia is also one of the clinical criteria used in the diagnosis of gout; however, elevated serum uric acid levels do not always lead to crystal deposition. Serum urate levels above 6.8 mg/dL can lead to the precipitation and accumulation of urate crystals in the joints and soft tissues, but acute gout attacks can occur even in patients with normal serum urate levels—and in these cases, the clinical diagnosis is more difficult and may be aided by imaging methods. Imaging studies can also be used in atypical cases, involving unusual ages or locations, with prolonged and less intense symptoms at the time of presentation.^{1,2}

Aware of what has been explained and reiterated in this case, even though the aforementioned osteoarticular conditions are amenable to evaluation through imaging examinations, regular follow-up of the patient by PHC would mitigate the adverse outcome of the condition. In this report, evaluations with higher technological density, such as optical microscopy and imaging examinations, play a secondary role compared to the potential for resolution offered by PHC in the follow-up of patients with gout.

Therefore, not only a rapid diagnosis and hypouricemic therapy are necessary, since the comprehensive approach, a core competence of the family and community physician, includes the management of comorbidities and the estimation of global cardiovascular risk and renal function. The risk of developing myeloproliferative or lymphoproliferative diseases also needs to be considered.¹²

In summary, the case report above highlights family and community medicine as a non-specialized field, encouraging the academic community to share successful experiences, especially in the context of PHC. In addition to the benefits described throughout this paper, it also adds the potential for reducing negative outcomes and hospitalizations due to chronic non-communicable diseases – which, in their main examples, are responsible for more than 80% of annual deaths worldwide.^{13,14}

CONCLUSION

In this case study, we see a 56-year-old male patient with examination findings of tophaceous lesions and nodules, particularly in the wrists, metacarpophalangeal joints, Achilles tendon, knees, and ankles,

consistent with the chronicity of gout. This report aims to raise awareness of the consequences of poorly controlled hyperuricemia in the long term and its impact on the natural history of the disease.

Beyond the clinical focus, this case report is expected to highlight the importance of coordinated care in healthcare practice. This work aims to strengthen the role of PHC as the preferred entry point for patients seeking longitudinal care, with non-communicable chronic diseases being one of its most important areas of focus.

Because it is a pathology that leads to serious conditions and complications, such as the renal insufficiency seen in this case, not only should pharmacological treatment be prescribed but also dietary and educational interventions for control. In the approach to the patient within the context of PHC in the Brazilian Unified Health System (SUS), equitable access to quality health services and comprehensive information about their condition is crucial.

Therefore, this case report reinforces the importance of an integrated approach to health in the early diagnosis, appropriate management, and prevention of complications in patients with gout, emphasizing the need for a comprehensive and integrated approach within the public health system.

The management of complex cases, whether from a clinical, demographic, or socioeconomic point of view, so prevalent in family and community medicine practice, reinforces the importance of continuity of care and coordination of care in the practice of general practitioners and the multidisciplinary team as a whole.

CONFLICT OF INTERESTS

Nothing to declare.

AUTHORS' CONTRIBUTIONS

JGRQ: Conceptualization, Supervision, Data curation, Formal analysis. MFCS: Conceptualization, Supervision. GGZ: Writing – review & editing. PAMF: Writing – original draft. LLF: Writing – original draft. AMF: Writing – review & editing.

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