BACKGROUND

Charcot-Marie-Tooth Disease (CMT) is the most common hereditary neuropathy, with an estimated prevalence of 40 cases per 100,000 individuals. CMT type 2, the most common subtype, is an axonal disorder, usually beginning during the second or third decade of life. Clinical features include distal weakness, muscle atrophy, reduced sensitivity, decreased deep tendon reflexes and deformity of the foot. It may lead to the development of Charcot arthropathy, with increased osteoclastic activity and joint destruction. Ultrasonography can show effusion, synovitis, high-grade doppler activity and bone irregularities, findings that can lead to a misdiagnosis of inflammatory arthritis.

CASE REPORT

Male, 27-year-old, reported a history of pain and warm swelling of the left ankle and midfoot, worsening after exercise and improving with rest, that started nine years ago and persisted for one year. During this period, he self-medicated with non-steroidal anti-inflammatory drugs. He denied associated symptoms such as ocular inflammation, skin lesions, diarrhea, urethritis or fever. Ten months ago, he began to present similar symptoms in right ankle and midfoot, after a 7-year asymptomatic period. He was referred to our Rheumatologic center for evaluation of oligoarthritis. He referred episodes of unnoticed trauma in lower limbs, only identified due to injuries and bleeding in pododactyls. He denied history of alcoholism or smoking. He mentioned that second-degree cousins had similar symptoms, without an etiologic diagnosis. Physical examination showed hammertoes, edema and local warmth in right midfoot, and limited left ankle range of motion. Glycemic and thyroid profile, vitamin B12 dosage and inflammatory tests were normal. Serologies for HIV, viral hepatitis and syphilis were negative. Radiograph of the left foot showed ankylosing tarsitis. Resonance of the right foot evidenced extensive synovitis associated with erosions/bone destruction with fragmentation and sclerosis involving the midfoot suggesting Charcot’s neuropathy. Electromyography was performed and showed motor and sensory axonal polyneuropathy in lower limbs compatible with Charcot-Marrie-Tooth type 2. Alendronate was prescribed to reduce bone turnover.

CONCLUSION

Charcot Arthropathy can cause joint effusion, synovitis, bone erosions and high-grade doppler activity at ultrasound examination. Neuropathic arthropathy should be suspected in patient with slowly progressive symptoms with distal predominance, weakness, sensory deficits, muscular atrophy and deformity of the feet. Treatment involves rehabilitation programs to prevent complications and increase functionality. Some small studies suggest that bisphosphonates may be beneficial in the acute phase because they reduce bone turnover and inflammatory process and delay the progression to joint deformity.