



Letter

A unique case of peroneus brevis/longus myositis in a patient with a STAT3 mutation

STAT3 mutations occur in patients with hyper-immunoglobulin E (IgE) syndrome (HIES). These patients experience recurrent infections involving opportunistic organisms of the lung and skin, including staphylococcal abscesses and pneumonias with pneumatoceles.^{1,2} The syndrome is also characterized by markedly elevated serum IgE levels and may include other features such as joint hyperextensibility, scoliosis, osteopenia, delayed shedding of primary teeth, coarse facial features, and pruritic dermatitis.^{3–5} The National Institutes of Health has developed a scoring system to classify patients with suspected HIES, with a score of 40 or higher being suggestive of HIES.²

We present a 36-year-old African American man with the history of a STAT3 mutation and a history of severe infections, including an empyema requiring thoracotomy with surgical lobectomy at age 2 years, knee abscess requiring incision and drainage, right arm abscess requiring incision and drainage, and four pneumonias requiring hospitalization. The patient's total serum IgE was 2,728 Ku/L, and his HIES National Institutes of Health score was 53, with points for his IgE level, skin abscesses, pneumonias pneumatocele, fractures, characteristic facies, eczema, upper respiratory infections, candidiasis, hyperextensibility, and increased nasal bridge. Genotyping showed a STAT3 mutation involving exon 12, Thr389Ile, which is consistent with the diagnosis of HIES.

He presented to the office complaining of persistent right lower leg pain with swelling for the previous 3 days involving the outer aspect of his right ankle. Despite the pain and swelling, he continued to ambulate without difficulty. The patient noted no history of trauma or repetitive movement to the foot or ankle and had no prior surgery or injury to that area. He did admit to scratching his foot and the development of excoriated areas of his forefoot before the symptoms. Consistent with his presentation of prior severe infections, he did not have fever, chills, or night sweats, but was requiring oral narcotics around the clock to continue his work.

The area of involvement of his right lower leg showed no warmth or erythema, although it was painful to manipulation. A magnetic resonance imaging (MRI) scan revealed inflammation of the peroneus brevis and longus muscle bellies (Fig. 1), and a serum creatine phosphokinase was elevated at 813, suggesting muscle involvement. Because of the patient's underlying immune status, he was referred to an infectious disease specialist who reviewed the MRI and started the patient on intravenous vancomycin and cefepime via peripherally inserted central catheter line for presumed bacterial myositis confined to the bellies of the peronius muscles. The patient's symptoms and clinical presentation improved dramatically within 24 hours of initiation of the antimicrobial treatment, and he completed his therapy with a course of oral

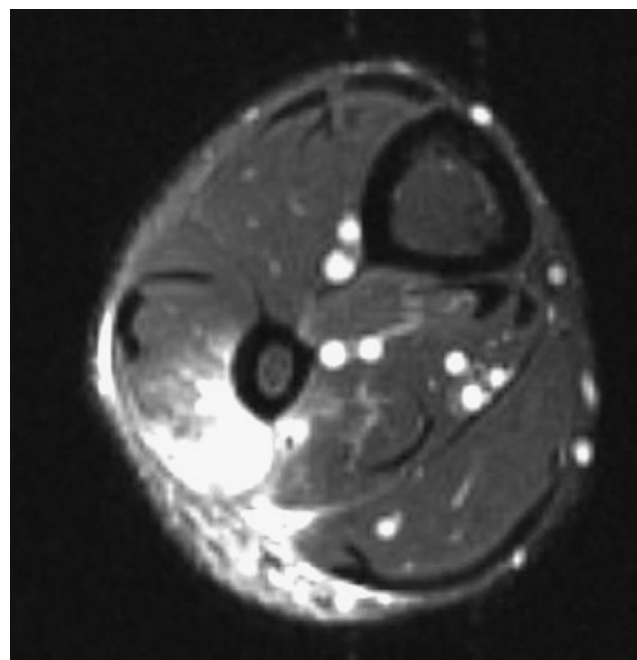


Figure 1. Cross-section of patient's MRI showing extensive enhancement of peroneus brevis and longus muscle bellies on T2 images.

antibiotics. Despite the patient's initial and subsequent complete blood counts failing to demonstrate leukocytosis, and negative blood cultures, a bacterial infection of the muscle bellies that stemmed from small excoriations located on the toes on the same (right) side was strongly suspected.

STAT3 modulates the expression of several genes that control central cell processes such as growth and death.⁶ STAT3 mutations have been associated with recurrent infections involving the skin, liver, and lung; however, isolated infections of individual muscles have not been reported.^{3,4} We report an unusual myositis of the peroneus longus and brevis muscles in a patient with a STAT3 mutation and HIES. This case demonstrates the necessity of high clinical suspicion of bacterial infection in evaluating patients with immunodeficiency, because patients with HIES may not demonstrate obvious clinical signs of infection.⁶

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- [2] National Institutes of Health. Table 1. *Scoring system* with clinical and laboratory tests for individuals in kindreds with *HIES*. Available at: http://www.niaid.nih.gov/LabsAndResources/labs/aboutlabs/lcid/stat3base/Documents/scoring_system.pdf. Accessed September 1, 2012.
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