Answer: Familial Mediterranean fever (FMF)

Discussion:

FMF is a hereditary autoinflammatory disease characterized by recurrent fevers and serositis as manifested by abdominal and chest pain, and synovitis as joint pain. Menstruation can be associated with FMF attacks. Erysipelas-like erythema (ELE) is reported in 7–40% of cases of FMF. ELE typically presents as a tender rash about the size of a palm occurring on the lower extremities and fades within 24–72 hours. ELE is the most common cutaneous manifestation of FMF, and physicians should consider FMF when patients have recurrent episodes of ELE. The differential diagnosis includes cellulitis, scarlet fever, and SLE. Cellulitis is a skin infection characterized by erythema, local tenderness, swelling and rubor. Because cellulitis is a bacterial infection, it does not match the recurrent and self-limited time course in this case. While 80–90% of patients with SLE develop skin lesions, a butterfly rash (a malar distribution over the cheeks and nose), photosensitivity, or discoid lesions are more common. The ANA test is positive in 95–100% of patients with SLE. Scarlet fever typically occurs in childhood and presents as a diffuse erythematous rash with numerous small papular elevations, commonly referred to as a “sandpaper rash.”

In our case, genetic testing revealed mutations in the MEFV gene (E148Q/P369S/R408Q), suggesting familial Mediterranean fever (FMF). After treatment with colchicine (1.0 mg/day), her symptomatic episodes ceased. Though we did not detect any signs of typical peritonitis, her recurrent abdominal pain and favorable response to colchicine were consistent with the Tel-Hashomer criteria for diagnosis of FMF.