Consultant360 Multidisciplinary Medical Information Network

CASE IN POINT

PEER REVIEWED

Familial Mediterranean Fever

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A 14-year-old girl of Turkish origin came to the clinic with concern for recurrent significant left knee, abdominal, and chest pain, along with fever lasting for approximately 2 days at a time and occasional eye pain. She denied aggravating factors, and her family history was noncontributory.

Physical examination. The patient was alert at presentation, with normal memory and intact cranial nerves function. The patient's blood pressure was 102/65 mm Hg, pulse was 77 beats/min, and temperature was 39.4°C. Her height was 150 cm and her weight was 42.64 kg, corresponding to a body mass index of 18.95 kg/m^2. Bowels sounds were normal, and the abdomen was nontender and nondistended. Lungs were clear to auscultation, and the heart rate and rhythm were regular without murmurs, rubs, or gallops. She reported having pain in the left knee, and an abdominal rash and generalized pain were present.

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Diagnostic tests. Clinical laboratory values were normal, except for positive results for antinuclear antibodies and anti-F-actin antibodies, with the rest of the lupus panel results being negative. Furthermore, the erythrocyte sedimentation rate was elevated at 29 mm/h (reference value, ≤20 mm/h or less), the C-reactive protein level was elevated at 3.93 mg/L (reference value, <0.80 mg/L), and the 25-hydroxyvitamin D level was low at 17 ng/mL. No other signs of active collagen vascular disease were present.

A clinical diagnosis of familial Mediterranean fever (FMF) was made based on the Tel-Hashomer criteria as described below.

Discussion. FMF is an autosomal recessive autoinflammatory disease that mainly affects the population of various Mediterranean ethnic groups. Characteristic features of this disorder include recurrent fever, monoarthritis, peritonitis, and pleuritis, with flares lasting for few days to weeks.[1] It affects the male and female populations equally; however, a slight male predominance has been reported.[2]

Mutations in the Mediterranean fever gene, *MEFV*, which encodes pyrin/marenostrin on the short arm of chromosome 16 (exons 2, 3, 5, and 10), have been implicated in the disease etiology.[3] More than 50 mutations associated with FMF have been detected; some mutations can cause more severe symptoms than others.[4]

FMF is subdivided into types 1 and 2. FMF type 1 is characterized by recurrent fever accompanied by pain and inflammation in parts of the body, while in type 2, amyloidosis is the first clinical manifestation.[5] The diagnosis is clinical and is based on the history of recurrent attacks and being in an ethnic group that FMF is known to affect. The Tel-Hashomer major and minor criteria and the Livneh simplified guidelines are often used to make the diagnosis.[6] Since our patient was of Turkish ethnicity; had recurrent febrile episodes; had abdominal, chest, and joint pain; and had a favorable response to colchicine treatment, she met all of the current criteria for FMF diagnosis.[7]

It is important to monitor the kidney, liver, and gastrointestinal tract closely in patients with FMF to monitor for and prevent the development of amyloidosis. Management includes treatment with nonsteroidal anti-inflammatory drugs and colchicine; recently, anakinra[8] and canakinumab[9] have shown great success in clinical trials.

Our patient is currently on colchicine, 0.6 mg orally twice a day; vitamin D_2, 50,000-IU capsule orally daily; D_3; 2000-IU capsule orally daily; and prednisone, 5 mg, 8 tablets daily orally, tapered, as needed. She is doing well and is being monitored on an as-needed basis.

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