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Case Report

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A Case of FMF that presents with epigastric abdominal pain and high amylase lipase: From suspicion to diagnosis of FMF

Abstract
Familial Mediterranean Fever is an autoinflammatory genetic disease usually seen in people of Mediterranean origin and characterized by attacks of fever and painful inflammation, especially in the abdomen, lungs, and joints. FMF patients presenting with abdominal pain may have signs of peritonitis, and thus the disease may mimic other acute abdominal conditions. This article presents a 21-years old male patient who was thought to have acute pancreatitis in the first evaluation but was finally diagnosed with FMF after more detailed anamnesis and further examinations. In young patients who present with abdominal pain and do not have a clear etiology, the diagnosis of FMF should also be considered.

Keywords: Abdominal Pain; Familial Mediterranean Fever; Acute Pancreatitis

INTRODUCTION

Familial Mediterranean Fever (FMF) primarily affects a population that originates from around the Mediterranean Sea and is characterized by peritonitis, pleuritis, arthritis, or erysipelas-like skin disease accompanied with recurrent episodes(attacks) of fever (1). Diagnosis of FMF is made based on clinical symptoms, taking into account the physical examination and laboratory findings with a careful anamnesis and the support of ethnicity and family history (2). The cause of abdominal pain seen in Familial Mediterranean Fever is inflammation in the peritoneum; therefore, patients may have peritonitis signs, and with these findings, the disease may be confused with other acute abdominal conditions.

CASE REPORT

A 21-years old male patient with known thalassemia minor and with no other chronic diseases, was admitted to our hospital with abdominal pain and fever complaints. The patient stated that the abdominal pain started the day before and gradually increased; it was not belt-like, did not hit the back, and did not change with movement. The patient did not describe nausea, vomiting, diarrhea, or constipation. The patient had no history of gallstones, alcohol use, medical drug use, and herbal or synthetic substance use. On physical examination, the patient's vitals were stable, conscious, oriented, and cooperative. In his abdominal examination, there was tenderness on superficial palpation in the epigastric region of the abdomen and tenderness on deep palpation in all quadrants of the abdomen, there was no palpable mass in the abdominal examination, and there was no guarding or rebound, palpable hepatomegaly and splenomegaly were not detected. There was no pathology on the respiratory system, cardiovascular system, neurological and musculoskeletal system. No rash and no palpable lymphadenopathy were detected on the patient's body.

The blood tests results of the patient were as follows; (WBC): 14.000\ml, neutrophils: 9700\ml, lymphocytes: 2000\ml, monocytes: 2000\ml, erythrocyte count (RBC):6,39 million/uL, hemoglobin: 11.8 g/dL, hematocrit: \%36, MCV (mean cell volume): 56 fL, c-reactive protein(CRP): 117 mg/L, amylase: 290 U/L, lipase: 255 U/L, glucose: 106 mg/L, urea: 19 mg/L, creatinine:0,86 mg/dL, ALT (Alanine Aminotransferase):14 U/L, AST (Aspartate Aminotransferase):21 U/L, LDH (Lactate Dehydrogenase):236 U/L. In Complete Urinalysis hemoglobin: (1+), leukocyte esterase (-), nitrite (-), protein (-), glucose (-), ketone (-), were detected. Acute pancreatitis was considered in the preliminary diagnosis of the patient who presented with

epigastric abdominal pain, and had elevated amylase and lipase levels. However, the diagnosis of acute pancreatitis was later dismissed because the patient's abdominal pain was not typical, and there were no findings in favor of pancreatitis or other abdominal pathologies in the Abdominal Computed Tomography (CT) imaging. Blood cultures and urine cultures were obtained in terms of a possible infection focus that may be present in the patient, and ceftriaxone antibiotic therapy was started empirically. A detailed physical examination of the patient has performed again, but no focus was observed to indicate infection. No infiltration was also detected in the thorax CT imaging.

Leukocyte esterase and other infection markers were found to be negative when the complete urinalysis was repeated. Infection disease department's consultation about the patient was also taken, and HIV (Human Immunodeficiency Virus), Hepatitis, CMV (Cytomegalovirus), Herpes, Rubella, EBV (Epstein Barre Virus), Mumps and Brucella tests were sent from the patient. Peripheral smear test of the patient was examined, erythrocytes were hypochromic microcytic, abundant rod cells were observed, platelets were sufficient, no atypical cells were observed. When the patient was questioned again in detail on the second day of his hospitalization, it was learned that the patient had similar complaints before, he had attacks in the form of abdominal pain and fever, and these attacks lasted for a few days. When the patient's family history was questioned, it was learned that his cousin had similar complaints. Thereupon, the diagnosis of Familial Mediterranean Fever (FMF) was considered in the patient. In addition to previous tests, ESR (Erythrocyte sedimentation rate) and fibrinogen tests were requested from the patient: ESR 29 mm/hr and fibrinogen 659 mg/dL. Thus, the patient were diagnosed with FMF regarding his abdominal pain, fever of 39 degrees, elevated white blood cell count, elevations in sedimentation, CRP, and fibrinogen levels and considering his abdominal pain and fever histories in the form of attacks, and colchicine treatment has started. After colchicine treatment, the patient's complaints regressed, and the inflammatory parameters returned to normal. Blood and urine cultures remained negative in the follow-up. Viral tests were negative, and brucella was negative, Eliza panel was negative. In the abdominal USG of the patient, bilateral kidney size, parenchymal thickness, and parenchymal echogenicity were reported as natural. In addition, FMF/MEFV gene target region/mutation analysis was requested from the patient for genetic counseling. The patient, whose complaints regressed and laboratory findings returned to normal, was discharged and called for a follow-up due to colchicine treatment and FMF gene mutation.

DISCUSSION

Acute pancreatitis (AP) is a clinical condition with local and/or systemic symptoms and signs due to inflammation secondary to local enzymatic activation in the pancreas. Patients with AP typically present with mid-epigastric and/or right upper quadrant pain; the character of the pain is generally continuous, knife-like pain and reflecting the back or sides. The diagnosis of AP is determined by the presence of typical abdominal pain, serum amylase and/or lipase values greater than three times the upper limit of normal, characteristic findings on abdominal imaging, and the presence of at least two of the criteria (3,4).

In the case we presented, our patient also applied with the complaint of abdominal pain in the epigastric region, but when the patient's pain was questioned in detail, it was not in the form of typical pancreatitis pain. Also, despite the patient's blood tests showed elevated amylase and lipase levels, the amylase and lipase elevations were not more than three times the reference upper limit, and no finding in favor of pancreatitis was reported in the abdominal CT (computed tomography) imaging. So, in the patient who was thought to have acute pancreatitis in the initial evaluation, the diagnosis of acute pancreatitis was dismissed.

Familial Mediterranean Fever (FMF) is an inherent disease characterized by signs of serosal inflammation such as abdominal pain, pleuritis, arthritis, and erysipelas-like skin lesions accompanied by recurrent episodes of fever (attacks). Familial Mediterranean fever (FMF) is generally considered an autosomal recessive disease, and affected individuals have biallelic pathogenic mutations in the MEFV gene located on the short arm of chromosome 16 (5,6). The disease is an inherited disease that usually occurs in people of Mediterranean descent, but it can affect people of any ethnic group (7). FMF diagnosis is made by careful anamnesis, clinical findings, family history, biochemical data, response to treatment, and exclusion of other familial periodic fever syndromes and supported by family history and ethnicity. The genetic tests for FMF are used to support diagnosis and consultation for family members in a patient who has clinic criteria of FMF (2).

The diagnosis of familial Mediterranean fever (FMF) is based on Tel-Hashomer clinical criteria, and the diagnosis of FMF is made in a patient with typical attacks using a combination of the following criteria: ≥1 major criterion or ≥2 minor criteria or 1 minor plus 5 supporting criteria or 1 minor criterion plus ≥4 of the first 5 supporting criteria (8).

Typical attacks are defined as episodes of pain associated with serositis, the presence of recurrent episodes (≥3 of the same type), the presence of fever (rectally measured fever of 38°C or higher), and short duration (12 hours to 3 days). Solitary episodes of fever can be considered a typical episode if they are recurrent, short-lived, and have no other definable cause (8).The major criteria used in the clinical diagnosis of FMF are peritonitis (generalized), pleuritic (unilateral) or pericarditis, monoarthritis (hip, knee, ankle), or fever alone, accompanying a typical attack. The minor criteria used in the clinical diagnosis of FMF are abdominal pain, chest pain, monoarthritis, exercise-related leg pain, and favorable response to colchicine. The supportive criteria used in the clinical diagnosis of FMF are family history of FMF, appropriate ethnic origin, age <20 years at onset of disease, an attack that is severe and requiring bed rest, attack's spontaneous remission, the symptom-free interval between

attacks, increase in inflammatory markers such as white blood cell count, sedimentation, fibrinogen, serum amyloid A within attacks, episodic proteinuria/ hematuria, history of negative laparotomy or a removal of the normal appendix, a history of consanguineous marriage (8).

In the case that we presented, our patient's attacks were typical. In addition, 1 major criterion was; peritonitis findings in our patient, 1 minor criterion was; favorable response to colchicine, 6 supportive criteria were; family history of FMF, Mediterranean origin, an attack that required bed rest, attack's spontaneous remission, the symptom-free interval between attacks and increase in inflammatory markers within attacks, Thus, FMF was diagnosed according to Tel-Hashomer clinical criteria.

As a result, the patient who applied with the complaint of epigastric abdominal pain and had high levels of amylase and lipase got diagnosed with FMF after deepening the patient's anamnesis and further examinations, although it was thought to might be acute pancreatitis in the first evaluation. In this case report, we wanted to draw attention to the fact that FMF can mimic other acute abdominal conditions such as acute pancreatitis clinically and as per laboratorial results, so the diagnosis of FMF should be kept in mind in young patients who presents with abdominal pain and do not have a clear etiology after the initial clinical, laboratory and radiological evaluations.

Conflict of interests
The authors declare that they have no competing interests.

Financial Disclosure
All authors declare no financial support.

Informed Consent
Written consent was obtained from the patient and his parents..

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