**Introduction**

- Familial Mediterranean fever (FMF) is an autoinflammatory disease characterized by relapsing and remitting episodes of fever and serositis.
- FMF is a clinical diagnosis but needs extensive workup to rule out other differentials.
- We describe a case of a 35-year-old male who presented with nausea, abdominal pain and myalgia found to have familial Mediterranean fever.

**Case**

A 35-year-old male presented with episodes of nausea, abdominal pain, and myalgia, lasting 1-4 days, every 2-5 weeks for almost a year.

**PMHx:** insulin dependent diabetes mellitus

**Physical exam:** mild diffuse abdominal tenderness

**Labs/ER visit:** Hb: 13.5 g/dL Cr: 0.80 mg/dL, HCO3: 22 mmol/L, blood glucose: 160 mg/dL, beta-hydroxybutyrate 11.5 mmol/L, and urine ketones 15 (1+).

**Gastrointestinal (GI) workup:** normal tissue transglutaminase IgA levels, endoscopy, colonoscopy and gastric emptying study.

**Rheumatology workup:** normal ESR, CRP, ANA, RF, cyclic citrullinated peptide (CCP) antibodies and Lyme screen.

**Hematology workup:** Normal IgA, IgM and IgG levels. Porphyria and hematopoietic malignancy workup was negative.

**Genetic testing:** found to have heterozygous V726A mutation for the MEFV gene.

**Treatment:** He was started on colchicine 1.2 mg daily with improvement of symptoms.

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**Approach to Familial Mediterranean Fever**

Differential diagnosis and workup for abdominal pain is broad and can be categorized systematically.

**Endocrinology:** Diabetes, Thyroid disease, Ketosis

**GI:** Celiac disease, Gastroparesis, Structural

**Rheumatology:** Lupus, Lyme disease and RA

**Hematology:** Multiple myeloma, Immunodeficiency, Porphyria

**Nutritional:** Vitamins deficiency, Anemia

**Diagnosis of FMF:**

- FMF is a clinical diagnosis presenting with episodic nature of symptoms.
- Family history and ethnicity of the disease can raise suspicion for FMF.
- The Tel Hashomer Medical Center criteria is widely used for diagnosis.
- Genetic testing for MEFV can help confirm the diagnosis.
- Response to colchicine further supports a diagnosis of FMF.

**Discussion**

- FMF is an auto inflammatory disease that mostly presents with fever and abdominal pain.
- It is more common in the Middle East and Eastern Europe, presenting mostly in patients < 20 years.
- Five gain-of-function mutations, V726A, M694V, M694I, M680I, and E148Q for the MEFV gene have been found which encodes for pyrin protein, resulting in a proinflammatory state.
- FMF is usually inherited as an autosomal recessive trait but some heterozygotes may also be symptomatic.
- Complications are secondary amyloidosis, small bowel obstruction and infertility.
- Treatment is colchicine but anti-inflammatory biologics (entanercept and canakinumab) can be used in refractory cases.

**Conclusion**

- Familial Mediterranean Fever can often present with recurring and relapsing fever and abdominal pain.
- In patients in which the etiology of abdominal pain remains uncertain, in the right clinical setting FMF may be considered.

**References**