Not the usual abdominal pain – a case of Familial Mediterranean Fever

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Introduction

- Familial Mediterranean fever (FMF) is an auto inflammatory 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.

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.

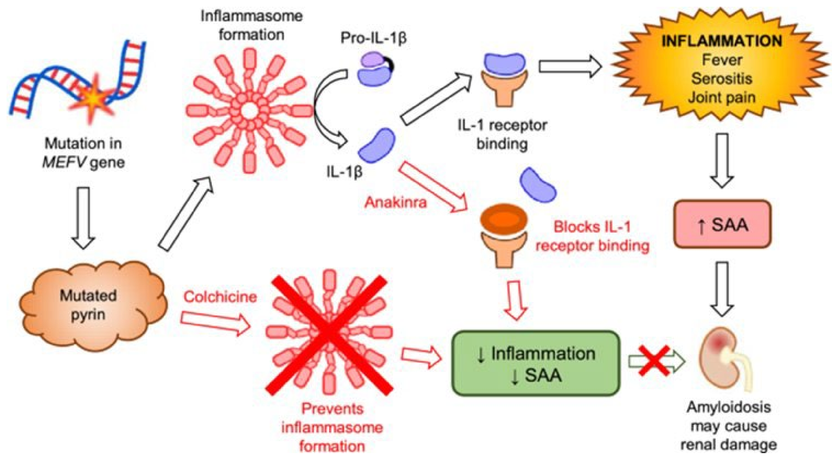

Figure 1: Pathophysiology of FMF

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 (entarancept 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

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