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Beyond the Mediterranean: Familial Mediterranean Fever Presenting as Recurrent Acute Abdomen in a South Asian Patient

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Abstract

Familial Mediterranean fever (FMF) is the most common autoinflammatory disorder and is characterized by recurrent, self-limited episodes of fever and serosal inflammation. It is most commonly described among individuals of Eastern Mediterranean and Middle Eastern ancestry; in other ethnic groups, FMF may be underdiagnosed due to its rarity and limited awareness. We describe a case of adult-onset FMF presenting with recurrent acute abdomen and mesenteritis in a South Asian man. The recurrent nature of the disease, marked systemic inflammatory response, radiologic evidence of mesenteric inflammation, and absence of mucosal disease on bidirectional endoscopies raised the possibility of an autoinflammatory disorder. An extensive evaluation for other inflammatory and autoimmune disorders was unrevealing. Further history revealed that several family members had experienced similar unexplained episodes. He was treated empirically with colchicine while awaiting genetic test results, with a favorable clinical response. Genetic testing later demonstrated compound-heterozygous MEFV variants, supporting the diagnosis. This case illustrates that FMF can occur outside its traditionally recognized ethnic and geographic populations; improved disease awareness and a high index of suspicion could avoid diagnostic delays. FMF should therefore remain a consideration in patients with recurrent, unexplained, self-limited febrile abdominal attacks, even in those without Mediterranean ancestry.

Categories: Emergency Medicine, Rheumatology, Gastroenterology
Keywords: acute abdomen, fever, mediterranean, mesenteritis, periodic

Introduction

Familial Mediterranean fever (FMF), also known as hereditary periodic fever, is the most common monogenic autoinflammatory disorder, characterized by recurrent, self-limited episodes of fever and serosal inflammation due to dysregulation of the innate immune system [1]. FMF follows an autosomal recessive inheritance pattern and results from pathogenic variants in the MEFV gene, which encodes pyrin [2]. FMF is most commonly reported among populations from the eastern Mediterranean and the Middle East, including Jews, Armenians, Turks, and Arabs. The classical presentation of FMF includes recurrent attacks of fever accompanied by serositis and large-joint arthritis. Acute abdominal pain is reported in as many as 90% of patients and is not infrequently mistaken for acute surgical abdominal illness [3]. FMF is diagnosed primarily clinically, with MEFV genetic testing providing additional support [4]. Colchicine remains the cornerstone of treatment, reducing recurrent attacks and substantially lowering the risk of sequelae [5].

We report a case of adult-onset FMF presenting with recurrent mesenteritis and abdominal inflammation in a person of South Asian descent. This case highlights the importance of considering FMF in the differential diagnosis of recurrent, unexplained abdominal inflammation, even in patients without a typical clinical presentation.

Case Presentation

A 46-year-old South Asian expatriate man initially presented to our institution in the United Arab Emirates in October 2024 with acute abdominal pain associated with fever. Computed tomography (CT) of the abdomen with contrast revealed concentric mural thickening of the sigmoid and descending colon with wall stratification, relatively engorged mesenteric vessels, and significant adjacent fat stranding. Fat stranding of the retroperitoneum at the root of the mesentery, mild thickening of the bilateral Gerota's and lateroconal fascia, and bilateral perinephric and paranephric fat stranding were noted as well. Laboratory investigations revealed leukocytosis and elevated C-reactive protein (CRP). Owing to concern for inflammatory bowel disease, the patient underwent endoscopic evaluation, with apparently normal visualized mucosa. No specific diagnosis was made during the initial presentation; however, he improved with symptomatic therapy and was discharged.

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In May 2025, during his vacation in Bangladesh, he had an episode of abdominal pain that was associated with a fever. Available records showed an inflammatory blood picture with diffuse left-sided colonic thickening, concerning for acute colitis. Blood and stool cultures, stool microscopy, and the Widal test were reported negative. He was treated for infectious colitis with ciprofloxacin and metronidazole. Symptoms resolved within 4-5 days of treatment.

Nearly two years after the initial episode, in July 2026, he presented again with acute onset of left-sided abdominal pain associated with fever, nausea, and vomiting. There were no associated symptoms such as diarrhea, overt gastrointestinal bleeding, weight loss, or jaundice. Contrast CT of the abdomen and pelvis reported significant mesenteric thickening with fat stranding and prominence of the mesenteric vessels around the small bowel loops in the central abdomen, predominantly on the right side (Figure *1*).

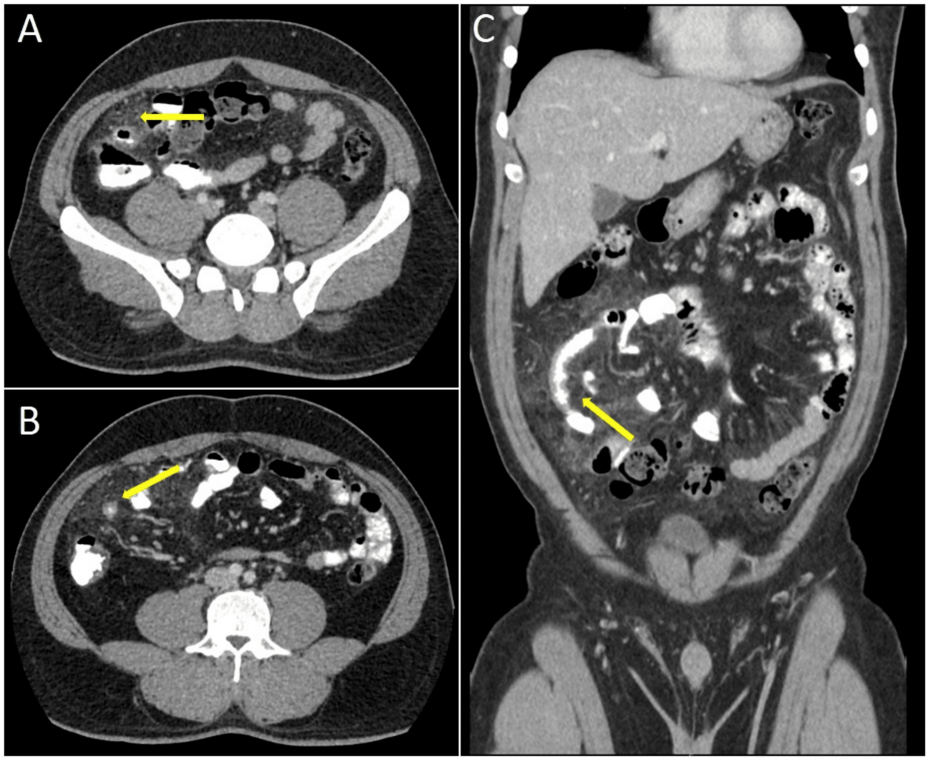


FIGURE 1: Axial (A and B) and coronal (C) computed tomography images of the abdomen, demonstrating fat stranding on the right side of the abdominal mesentery (yellow arrows), with thickening of adjacent small bowel loops

Multisegmental mural thickening of the small bowel loops was noted in the right central and lower abdomen, predominantly involving the ileal loops, along with mild mural thickening involving the ascending and descending colon. Laboratory investigations revealed an elevated leukocyte count, CRP, and ferritin (Table *1*). Antinuclear antibodies, anti-neutrophil cytoplasmic antibodies, immunoglobulin G4, and porphyrin levels were within normal limits. Bidirectional endoscopy and enteroscopy were performed but were unrevealing, including random mucosal biopsies. Symptoms resolved with symptomatic and supportive therapy in around three days.

Parameter	October 2024	May 2025	July 2026	Normal
Hemoglobin	14.3	13.9	14.1	13.5-17.5 g/dL
WBC	12.76 > 11.8	13.8 > 11.3	13.2 > 11.7	4.0-11.0 × 10³/µL
Platelet	365	432	486	150-450 × 10³/µL
CRP	150.6	117	287	<5 mg/L
ESR	25	-	37	0-15 mm/hour
Creatinine	0.8	0.9	0.8	0.7-1.3 mg/dL
Bilirubin	0.7	0.9	0.9	0.2-1.2 mg/dL
ALT	38	40	36	8-40 U/L
AST	32	41	39	10-40 U/L
Lactate	0.8	-	0.9	0.5-2.2 mmol/L
Blood culture	No growth	No growth	No growth	No growth
Ferritin	467	-	735	30-400 ng/mL
LDH	-	-	229	140-280 U/L
IgG4	-	-	54	4-86 mg/dL
ANA panel	Negative	-	Negative	Negative
ANCA panel	-	-	Negative	Negative
QuantiFERON	-	-	Negative	Negative
Widal	-	Negative	-	Negative
Dengue	-	Negative	-	Negative
Malaria	-	Negative	-	Negative
Stool analysis	-	No ova/parasites	-	No pathogens
Stool culture	-	No growth	-	No growth
Urinalysis		Normal (no proteinuria)		Normal

TABLE 1: Summary of the laboratory workup

WBC: white blood cell count, CRP: C-reactive protein, ESR: erythrocyte sedimentation rate, ALT: alanine aminotransferase, AST: aspartate aminotransferase, LDH: lactate dehydrogenase, IgG4: immunoglobulin G subclass 4, ANA: antinuclear antibody, ANCA: antineutrophil cytoplasmic antibody, "-": not available.

Recurrent episodes of febrile acute abdomen with elevated inflammatory markers and mesenteritis, without apparent endoscopic mucosal disease, raised suspicion for inflammatory or autoimmune diseases (autoinflammatory syndromes, IgG4-related mesenteritis, systemic vasculitis, small bowel Crohn's disease, etc.). A retrospective detailed review of the medical history revealed that multiple family members were suffering from similar clinical symptoms, without a definite diagnosis. Given the concern for FMF, blood samples were collected for MEFV gene mutation analysis, and he was started on empirical colchicine therapy, to which he responded. Subsequently, genetic testing was reported as positive for a compound-heterozygous MEFV mutation (E148Q/P369S), further confirming the diagnosis. He was recommended to undergo long-term colchicine therapy for FMF.

Discussion

Autoinflammatory syndromes are a class of disorders characterized by recurrent febrile illness associated with inflammatory mucosal, serosal, cutaneous, or articular manifestations; the prototype and most common is FMF [6]. Gain-of-function mutations involving the MEFV gene on chromosome 16 (16p13), which encodes the protein "pyrin," are the key genetic driver of this uncommon pyrexia syndrome. Abnormal activation of the pyrin inflammasome results in excessive IL-1β-mediated inflammation [2]. As the name

suggests, the disease is most prevalent in the Eastern Mediterranean region, with prevalence as high as 1 in 400 to 1000. The highest number of FMF cases are reported from Turkey, followed by Israel and Armenia [7]. Although population migration has contributed to its recognition and diagnosis outside these traditionally prevalent zones, it is rarely reported among South Asians.

Clinically, FMF is characterized by recurrent, short-lived attacks of fever, usually lasting 1-3 days, accompanied by peritonitis, pleuritis, or acute monoarthritis involving the large joints. Less common manifestations include erysipelas-like erythema, pericarditis, and protracted febrile myalgia [2]. Abdominal attacks occur in approximately 90% of patients and may resemble an acute surgical abdomen. During flares, patients typically develop a rapid neutrophilic acute-phase response, with clinical symptoms resolving and well-being restored between episodes [3]. The diagnosis of FMF is primarily clinical and supported by validated classification criteria, most notably the Tel-Hashomer/Livneh criteria (Table *2*), which incorporate characteristic attacks and response to colchicine and have reported sensitivity and specificity of approximately 95% and 97%, respectively. Genetic testing for MEFV, in isolation, is often supportive rather than diagnostic. Up to one-third of clinically diagnosed patients may have only a single detectable MEFV variant, and a negative or inconclusive result does not exclude FMF when the clinical phenotype and family history are strongly suggestive [8]. The newer Eurofever/PRINTO diagnostic criteria additionally incorporate genetic findings [3,5].

Category	Criteria
Major criteria	1. Typical attacks of generalized peritonitis
	2. Typical attacks of unilateral pleuritis or pericarditis
	3. Typical attacks of monoarthritis (hip, knee, or ankle)
	4. Typical attacks of fever alone
	5. Incomplete abdominal attacks
Minor criteria	1. Incomplete chest attacks
	2. Incomplete joint attacks
	3. Exertional leg pain
	4. Favorable response to colchicine
Supportive criteria	1. Family history of FMF
	2. Appropriate ethnic origin
	3. Age at disease onset <20 years
	4. Severe attacks requiring bed rest
	5. Spontaneous remission of attacks
	6. Symptom-free intervals
	7. Transient elevation of inflammatory markers
	8. Episodic proteinuria or hematuria
	9. Previous negative laparotomy or appendectomy
	10. Parental consanguinity
FMF diagnosis supported	≥1 major criterion, OR,
	≥2 minor criteria, OR,
	1 minor criterion + ≥5 supportive criteria

TABLE 2: Livneh criteria for familial Mediterranean fever (FMF)

Early recognition and treatment of FMF are important because persistent inflammation can lead to AA amyloidosis, which remains the most serious long-term complication of the disease. Renal involvement may present with proteinuria and progressive renal dysfunction, particularly in patients with prolonged disease

activity and persistent subclinical inflammation. Daily colchicine is the first-line treatment for FMF. It substantially reduces the frequency and severity of attacks and, most importantly, prevents AA amyloidosis, even among patients with incompletely controlled attacks. The therapeutic objective of long-term colchicine is to completely suppress subclinical inflammation [9]. Once- and twice-daily colchicine regimens have shown comparable efficacy, allowing dosing flexibility [9]. Approximately 10-15% of patients have an inadequate response or intolerance to colchicine and may require IL-1-targeted therapy with anakinra or canakinumab [10].

Our patient presented with recurrent, self-limited febrile episodes and acute abdominal pain, prompting diagnostic evaluation. At presentation, we did not even consider FMF. However, there was no definitive diagnosis to explain the patient's presentation, most likely due to a lack of awareness of the disease and the patient's ethnic descent, in which FMF is exceedingly rare. However, during the subsequent episode, the patient underwent a more extensive evaluation that culminated in the final diagnosis. The episodes were associated with a marked systemic inflammatory response and mesenteric fat stranding during the attacks. Extensive evaluation excluded infection, inflammatory bowel disease, systemic vasculitis, and IgG4-related disease. The family history, unequivocal response to colchicine, and an MEFV mutation confirmed the diagnosis. This case shows that undetected FMF kindreds exist among the South Asian population. An analysis of genetic pipelines from the 1000G and NHLBI datasets has reported MEFV mutations among Bengalis from Bangladesh and Punjabis from Lahore, Pakistan [11]. Our experience highlights the challenges of FMF diagnosis and calls for a structured approach based on clinical presentation, even in patients outside endemic areas, to enable timely diagnosis.

Conclusions

FMF is characterized by recurrent, self-limited febrile attacks with serosal or mesenteric inflammation, classically seen in individuals of Mediterranean ancestry. The diagnosis is often clinical, and genetic testing for MEFV supports it. The diagnosis is potentially challenging, especially in patients of non-Mediterranean ancestry with adult-onset symptoms. This case highlights the importance of maintaining a high index of suspicion for FMF to facilitate recognition of this frequently overlooked condition without delay in patients with recurrent, self-limited fever and abdominal pain. In patients from high-prevalence areas, and when suspicion is high, genetic testing may enable earlier recognition. Early initiation of colchicine is essential to prevent recurrent inflammatory episodes and reduce the risk of amyloidosis, the major long-term complication of FMF.

Additional Information

Author Contributions

All authors have reviewed the final version to be published and agreed to be accountable for all aspects of the work.

Acquisition, analysis, or interpretation of data: Mathew Vadukoot Lazar, Shruthi Chamala, Debashish Mishra

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Disclosures

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