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CASE REPORT

First Genetically Confirmed Case of Familial Mediterranean Fever in Somalia: A Case Report and Diagnostic Challenges in Resource-Limited Setting

Mohamed Abdikani Jama[1], Abdi Rizaq Hashi Hersi[2], Abdifetah Ibrahim Omar[2], Abdifatah Abdullahi Jalei[2,3], Burhan Abdullahi Kulmiye[2]

[1]Faculty of Medicine, Pediatric Residency Cairo university, Cairo, Egypt; [2]Faculty of Medicine and Health Science, Jamhuriya University of Science and Technology, Mogadishu, Somalia; [3]National Public Health Referral Laboratory, Ministry of Health and Human Services, Mogadishu, Somalia

Correspondence: Abdifetah Ibrahim Omar, Email abdifatah@just.edu.so

Abstract: Familial Mediterranean Fever (FMF) is a hereditary autoinflammatory disease characterized by recurrent fever, serositis, and systemic inflammation, primarily affecting Mediterranean populations. It is rarely reported in sub-Saharan Africa and remains underdiagnosed due to limited awareness and lack of genetic testing services. We report a 57-year-old Somali woman with a four-year history of recurrent high-grade fever, erysipelas-like erythema, and polyarthralgia. Due to the absence of diagnostic facilities in Somalia, she was referred to Egypt, where genetic testing confirmed an MEFV E148Q mutation. Colchicine and supportive therapy led to clinical improvement and reduced inflammatory markers. This is the first documented case of FMF in Somalia. There is an urgent need to improve physician awareness and expand local diagnostic capacity. In low-income settings like Somalia, where many people live in poverty, seeking medical care abroad is not feasible for most. Failure to address these gaps risks avoidable suffering, life-threatening complications, and worsening health inequities.

Keywords: familial Mediterranean fever, Somalia, high-grade fever, erysipelas-like erythema, polyarthralgia, p.E148Q

Introduction

Familial Mediterranean Fever (FMF 249100) is a hereditary autoinflammatory disorder that typically manifests in childhood or early adulthood.[1] It is characterized by recurrent episodes of fever, polyarthralgia, erythematous skin rashes, serositis, and abdominal pain.[2] If left untreated, FMF can lead to severe complications such as amyloid-associated (AA) amyloidosis and progressive kidney failure. The condition is caused by mutations in the MEFV gene which codes many variants like p.E148Q in exon 2 by Ashkenazi Jews 12, which encodes pyrin, a key protein involved in regulating the innate immune response. Genetic testing for MEFV mutations remains the standard diagnostic tool; a positive result confirms the diagnosis and guides management.[3]

FMF predominantly affects individuals of Mediterranean origin, including Armenians, Turks, Arabs, and North Africans. The highest prevalence is observed in Turkey, with rates of approximately 1 in 1,000 individuals.[4] In Africa, FMF displays diverse epidemiological and genetic profiles. North African countries, in particular, report high *MEFV* mutation carrier frequencies with marked geographic variability. For instance, the p.M694I mutation, often associated with amyloidosis, predominates in Algeria, while the p.V726A variant is more frequent in Tunisia and Morocco.[5] An estimated 8% carrier rate has been reported across Middle Eastern and North African populations.[6] These variations underscore the importance of region-specific diagnostic strategies, as genotype-phenotype correlations may affect clinical severity and outcomes.

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In sub-Saharan Africa, however, FMF remains significantly underdiagnosed. Contributing factors include the limited availability of genetic testing, lack of clinical awareness, and the overlap of FMF symptoms with common infectious diseases such as malaria and tuberculosis. A notable case from Tanzania described a 34-year-old Egyptian man with recurrent fever, abdominal pain, and pleuritis. He was diagnosed clinically using Livneh's criteria and responded well to colchicine treatment.[7] This highlights the diagnostic challenges faced in non-endemic and resource-constrained settings, where FMF may be overlooked despite clinical presentation.

To our knowledge, this is the first documented case of FMF in Somalia. The patient, an adult Somali woman, presented with recurrent high-grade fever, erysipelas-like erythema, and polyarthralgia that had persisted for over four years. Due to the rarity of FMF in this region and the non-specific nature of her symptoms, the diagnosis was delayed. The lack of molecular diagnostic facilities in Somalia prevented local confirmation of FMF. Consequently, the patient was referred to Egypt, where *MEFV* gene testing confirmed the diagnosis and used clinical criteria from Tel hashomer and Eurofever criteria and then patient colchicine therapy was initiated with clinical improvement. This case underscores the broader diagnostic limitations in Somalia and the potential for preventable complications such as amyloidosis and unnecessary surgeries when FMF remains unrecognized.

Case Report

A 57-year-old Somali woman presented with four-year history of recurring symptoms, initially including nausea, vomiting (2–3 times daily), fatigue, and increased urinary frequency. These symptoms were later followed by high grade fevers (39–40°C, axillary) and painful, highly sensitive erythema that first appeared on the right leg, and eventually spread to the breasts, becoming bilateral. These episodes were accompanied by arthralgia, myalgia, and headaches, typically lasting 3 to 7 days. Initially, occurring every 4–6 months, the frequency of attacks increased over time to once every 1–2 months in four years.

The patient sought care at multiple hospitals in Mogadishu, Somalia, where physicians suspected Familial Mediterranean Fever; however, due to the lack of genetic testing facilities in Somalia, she was advised to seek diagnosis abroad. Between February and April 2025, the patient continued to experience frequent episodes, and was eventually referred for further consultation and treatment in Egypt, where her symptoms were noted to worsen with emotional stress.

In Egypt, an internal medicine specialist ordered laboratory investigations. Genetic testing did polymerase chain reaction (PCR) revealed a pathogenic MEFV mutation at codon p.E148Q Heterozygous. Laboratory results showed markedly elevated markers: Serum Amyloid A (SAA) (68 mg/dL; ref. 0–0.6), C-reactive protein (CRP) (19.5 mg/dL, ref. 0–5), Erythrocyte Sedimentation Rate (ESR) (120 mm in the initial hour), and C3 complement (216 mg/dL, ref. 90–180). Protein/creatinine ratio was elevated at 852 mg/dL (ref.0–200), though creatinine remained normal at 0.530 mg/dL. Lipid panels showed triglycerides (202 mg/dL; ref. 0–150), Low-Density Lipoprotein (LDL) (143.6 mg/dL; ref. 0–100), Very Low-Density Lipoprotein (VLDL) (40 mg/dL; ref.0–30), and Non-High-Density Lipoprotein Cholesterol (184 mg/dL; ref. 0–130), finally urinalysis revealed no albumin and erysipelas like lesion was observed (Table 1, Figure 1).

Table 1 Laboratory Findings Supporting the Diagnosis of MEFV

Category	Test Name	Result	Unit	Reference Range
Genetic Screening	MEFV Gene (P.E148Q)	Positive heterozygous	–	Not Detected
	SAA	68.00 ↑	mg/L	0 – 6.4
Inflammatory Markers	CRP	19.5 ↑	mg/L	0 – 5
	ESR	120 ↑	mm	Up to 14
	Total Cholesterol	221 ↑	mg/dL	Up to 200
	Triglycerides	202 ↑	mg/dL	0 – 150
	HDL Cholesterol	37	mg/dL	≥60 (Low risk)

(*Continued*)

Table 1 (Continued).

Category	Test Name	Result	Unit	Reference Range
Lipid profile	LDL Cholesterol	143.6 ↑	mg/dL	0 – 100
	VLDL Cholesterol	40.4 ↑	mg/dL	0 – 30
	Non-HDL Cholesterol	184 ↑	mg/dL	0 – 130
	HDL Risk Factor	6	-	Mild risk: 4.4
	Color	Yellow	-	Yellow
	Aspect	Clear	-	Clear
	Volume	10.0	mL	4 – 8
	Reaction (pH)	6.0	-	-
Urine Analysis	Specific Gravity	1.015	-	1.005–1.025
	Nitrite,	Negative	-	Negative
	Albumin,			
	Sugar,			
	Acetone,			
	Bile salts			
	Bile Pigment			
	Urobilinogen	Normal Trace	-	-
	Pus Cells	3–5	/H.P.F	0 – 5
	RBCs	0–2	/H.P.F	0 – 5
	Ionized Calcium	1.20	mmol/L	1.12–1.32
	Serum Creatinine	0.530	mg/dL	0.5–1.1
Kidney Function Tests	Blood Urea Nitrogen (BUN)	12.0	mg/dL	6 – 20
	Serum Uric Acid	5.8 ↑	mg/dL	2.4–5.7
	Serum Calcium	10.4 ↑	mg/dL	8.6–10.2
Body Fluid Chemistry Analysis	Urinary Protein	64.78	Mg/dl	-
	Urinary Creatinine	76	Mg/dl	–
	Protein/Creatinine Ratio	852.37↑	Mg/Creatinine	0 – 200

Regarding imaging, Echocardiography showed preserved ejection fraction of 58% (normal range: 50–70%). Computed Tomography (CT) abdomen and pelvis were unremarkable. Color duplex imaging showed varicose vein in the right leg and massive saphenous vein inflammation in the left. Furthermore, Magnetic Resonance Imaging (MRI) of the left thigh showed subcutaneous swelling and minor bone and joints fluid changes (Figure 2). Finally, PET CT revealed inflammatory changes consistent with right hip (Figure 3).

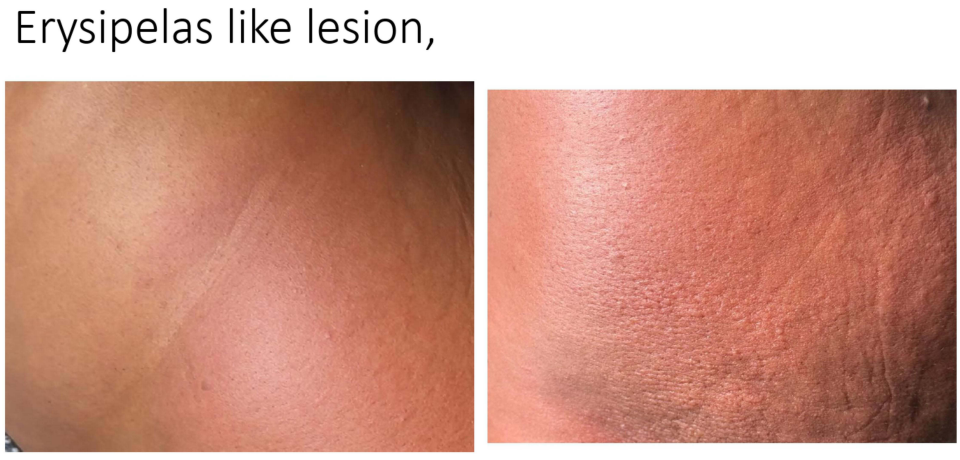

Figure 1 Erysipelas like lesion was observed at the diagnosis of FMF.

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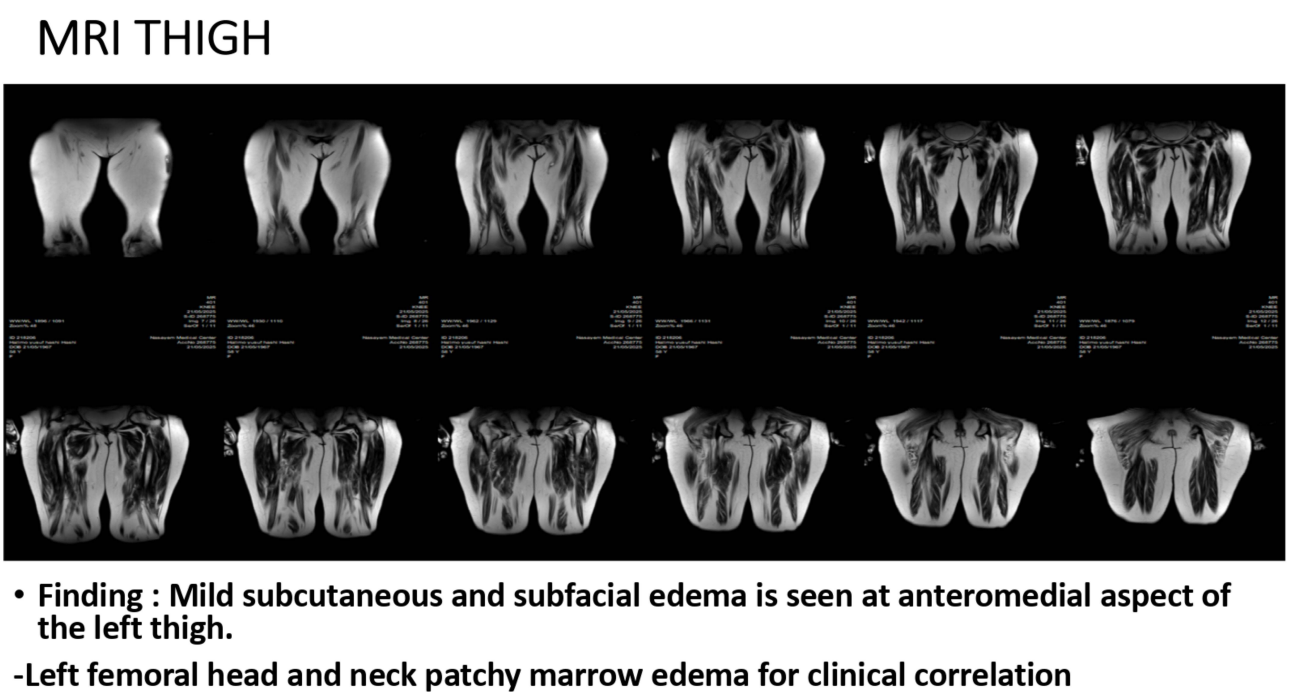

Figure 2 MRI of mild subcutaneous and subfacial edema is seen at anteromedial aspect of the left thigh.

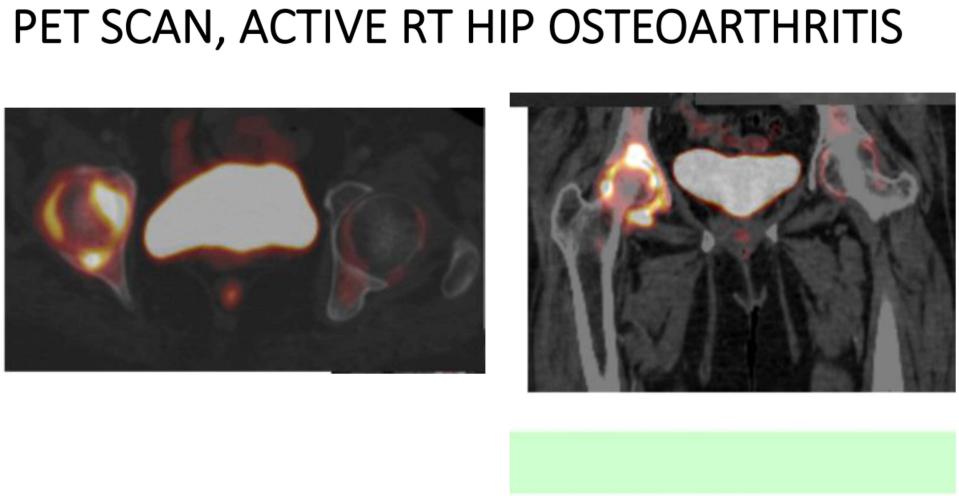

Figure 3 PET CT Scan of abdomen and pelvic showing right side Hip.

Treatment was initiated with 0.5 mg colchicine twice daily and prednisolone 20 mg, tapered to 10 mg over four weeks. The patient had received a clinical diagnosis prior to genetic confirmation, and improvement was observed within one week of colchicine initiation.

At the four-week follow-up after treatment initiation, the patient's ESR had decreased, but CRP and SAA levels remained elevated despite the colchicine regimen. Following a subsequent attack, she received an intramuscular (IM) injection of ketorolac tromethamine (30 mg/2 mL) and a combination tablet of diclofenac and methocarbamol (50 mg/500 mg), which provided greater symptom relief than colchicine alone (Table 2).

The elevated protein/creatinine ratio indicated potential renal dysfunction. Ramipril tablet 1.25 mg twice daily was initiated, leading to a marked reduction in protein/creatinine ratio to (91.14 mg/dL; ref. 0–200) within seven days. The

Table 2 Follow-Up During Acute FMF Attack After Initial Treatment with Colchicine (0.5 mg BID), Prednisolone, and Ramipril (1.25 mg BID)

Category	Test Name	Result	Unit	Reference Range	Previous Result
Body Fluid Chemistry Analysis	Urinary Protein	7.20	mg/dL	—	64.78
	Urinary Creatinine	79	mg/dL	—	76
	Protein/Creatinine Ratio	91.14	mg/g creatinine	0 – 200	852.37
Inflammatory Markers	ESR	90	mm	Up to 14	120
	SAA	>238.00	mg/L	0 – 6.4	68.00
	CRP	↑ 159.1	mg/L	0 – 5	19.5

Table 3 Follow-Up After Escalated Treatment with Colchicine (0.5 mg TID), Ramipril, Levofloxacin, Piroxicam (20 Mg), Ezetimibe (10 Mg), and Atorvastatin (20 Mg)

Category	Test Name	Result	Unit	Reference Range	Previous Result
Inflammatory Markers	SAA	52.1	mg/L	0 - 6.4	>238.00
	CRP	3.0	mg/L	0 - 5	159.1
	ESR	90	mm	Up to 14	90
Kidney Function Tests	Serum Creatinine	0.76	mg/dL	0.6–1.2	0.530
	Serum Uric Acid	4.3	mg/dL	2.6 - 6	5.8
Lipid Profile	Serum Total Cholesterol	167	mg/dL	Up to 200	221
	Serum Triglycerides	144	mg/dL	0 - 150	202
	HDL Cholesterol	51	mg/dL	≥ 60 (Low risk), < 40 (High risk)	37
	LDL Cholesterol	87	mg/dL	0 - 100	143.6
	VLDL Cholesterol	29	mg/dL	0 - 30	40.4
	Non-HDL Cholesterol	116	mg/dL	0 - 130	184
	HDL Risk Factor	3.27	-	Mild: 4.4, Mod: 7.1, High: 11.0	6
Body Fluid Chemistry	Urinary Protein	18.53	mg/dL	-	7.2
	Urinary Creatinine	143	mg/dL		79
	Protein/Creatinine Ratio	129.58	mg/g creatinine	0 - 200	91.14

dyslipidemia was addressed with ezetimibe 10 mg and atorvastatin 20 mg for 21 days. Based on a high HDL risk factor 6 (mild risk: 4.4; moderate: 7.1), the patient continued lipid lowering therapy under follow-up.

The patient has been administered 0.5 mg of colchicine three times daily to the patient. Clinical symptoms have greatly improved, although laboratory investigations have shown partial improvement. The amyloid levels remain elevated but have decreased compared to the previous result, while CRP has returned to normal; nevertheless, ESR is unchanged from the last test and has not yet reached normal levels. The patient vital signs are normal and healthy, and there is no sign of abdominal or joint pain. Colchicine has no side effect to the patient; therefore, the physician recommends continuing the treatment to prevent illness progression and FMF attacks. Additionally, physician educate patients about medication adherence, and importance of families in lifestyle support, and ensure consistent monitoring. The physician advises the patient to return after one month to assess inflammatory markers such as ESR, CRP, and amyloid, and to evaluate for any recurrence of symptoms (Table 3).

The Literature Review and Discussion

FMF is most prevalent among populations in the Mediterranean region, including Turkey, Greece, Arab populations, and Jewish communities.[6] The migration of individuals from these regions to Europe and North America has led to an increased recognition of FMF cases outside its traditional endemic nations. In contrast, FMF remains exceedingly rare and underreported in sub-Saharan Africa, particularly in countries such as Somalia, due to the lack of genetic testing, serum amyloid A facilities, and absence of family history.[5] This rarity often results in delayed diagnosis, with patients

frequently presenting at more advanced stages of the disease, thereby reducing the chances of optimal treatment outcomes and increasing the risk of complications.

The pathogenesis of FMF is linked to mutation in the MEFV gene, which encodes the pyrin protein. Pyrin plays crucial role in regulating the inflammatory response through the activation of interleukin-1beta (IL-1β), a key pro-inflammatory cytokine. Mutation in MEFV result in uncontrolled activation of pyrin, leading to recurrent episodes of inflammation.[8] Clinically, FMF is characterized by recurrent attacks of fever, serositis (abdominal pain), arthritis, and erysipelas-like skin rashes. These symptoms can be triggered by cold exposure, stress, or menstruation. Joint involvement often affects the hips, knees or ankles, and cutaneous manifestations typically present as erythematous, tender rashes on the lower extremities. The most severe complication of untreated or unrecognized FMF is secondary (AA) amyloidosis, predominantly affecting the kidneys and potentially progressing to nephrotic syndrome and end-stage renal disease, subsequently requiring dialysis or kidney transplantation.[9] In this patient, she carried p.E148Q mutation in the MEFV gene, a common variant in Mediterranean and North African populations. Its pathogenicity is often debated, as it is associated with milder phenotypes. Still, it is recognized in diagnostic criteria, and in symptomatic cases like ours, it holds clinical significance.

Although FMF usually begins in childhood or early adulthood,[10] this case is unusual, because the patient is 57-year-old woman. It is unclear whether her symptom began earlier in life and went unrecognized or if they truly began later. There is no clear evidence that gender and age significantly effects disease onset or severity. However, this case demonstrates that FMF can still occur in older age and should not be excluded based on age alone.

FMF can be diagnosed using various criteria, including Tel Hashomer, Yalcinkaya-Ozen, and Eurofever/PRINTO. Among these, the Tel Hashomer criteria, one of the most established and commonly used, require at least two major or one major and two minor criteria. Our patient meets three major (Recurrent febrile fever, Amyloidosis without predisposing disease, and our patient responded to colchicine) and two minor criteria (Recurrent febrile fever, and Erysipelas skin lesion). Additionally, the more recent Eurofever/PRINTO criteria, which incorporate both clinical (Arthritis and Abdominal pain) and genetic components (MEFV gene mutation), were also met in this case, further confirming the diagnosis of FMF. While Livneh's criteria are also widely used for clinical diagnosis, particularly in certain populations as noted in our introduction, our specific case was evaluated and diagnosed using the Tel Hashomer and Eurofever/PRINTO criteria.

Physicians often rely on a patient's medical history, family background, ethnicity, and physical examination to diagnose FMF. This patient presented with elevated inflammation markers including high levels of CRP, ESR, and white blood cells with neutrophilia, consistent with previous reports.[9] Although her urinalysis revealed proteinuria, a potential warning sign for kidney damage due to amyloidosis, her echocardiography and renal function tests were normal. The proteinuria resolved after treatment with an ACE inhibitor, suggesting a non-amyloid cause. The pattern of elevated inflammatory markers during attacks and normal levels between episodes is characteristic of FMF, and helps differentiate it from other inflammatory conditions.[9]

Colchicine remains the first-line treatment for FMF. It reduces the frequency and severity of attacks, improves quality of life, and prevents complications such as amyloidosis. The recommended dose is ≤0.5 mg/day for children under five years old (or ≤0.6 mg/day with 0.6 mg tablets), and 1.0–1.5 mg/day for adults (up to 1.8 mg/day with 0.6 mg tablets). Our patient is receiving 0.5 mg twice daily, which is within the standard adult dosage range.

Limitations of this case include the absence of comprehensive family history, which could have provided further insight into the genetic predisposition and penetrance of the E148Q mutation within her lineage. Broader genetic screening of her family members was also not feasible due to resource constraints. Furthermore, the follow-up period for this case was relatively short, and while clinical improvement was noted, certain biomarkers like ESR and serum amyloid A did not fully normalize, indicating a potential ongoing inflammatory burden or a need for optimized colchicine dosage. Access to advanced treatments, such as anti-IL-1 agents (biologics), was also unavailable in the local setting, limiting treatment escalation options beyond colchicine if the patient had proved refractory. These challenges highlight the significant barriers to optimal FMF management in resource-limited environments like Somalia.

Conclusion

This case may highlight the presence of FMF in Somalia and the critical gaps in diagnosing rare genetic disorders in low-resource settings. The patient's delayed diagnosis, primarily driven by limited clinical awareness and the absence of genetic testing, led to prolonged suffering and the need for costly treatment abroad. We acknowledge that the pathogenicity of p.E148Q is debated. The presence of a single variant of uncertain significance allele like E148Q alone may not definitely support an FM diagnosis. However, in conjunction with the patient's strong clinical presentations fulfilling both Tel- hashomer and Eurofever/PRINTO criteria and clear response to colchicine therapy, we believe that this case's unique genetic testing abroad significantly contributed to overall diagnosis of FMF in this unique setting.

Improving early recognition through clinician training, incorporating FMF into the differential diagnosis of recurrent fevers, and expanding access to diagnostic tools are essential steps. In a country where most people live in poverty, seeking medical care abroad is simply not an option for the majority. This deepens health inequities and leads to avoidable, life-threatening complications. Strengthening local diagnostic and treatment capacities is not just important, it is urgent, otherwise many patients will continue to suffer or die from conditions that are otherwise treatable.

We strongly advocate for regional or international collaborations to improve access to molecular diagnostics for FMF. Simultaneously, there is a pressing need to raise clinical awareness among primary care physicians across sub-Saharan Africa to consider FMF in recurrent fever case, particularly after excluding more common infectious diseases.

Ethical Approval

Ethical approval was not required for this case report.

Consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images.

Acknowledgments

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Author Contributions

All authors made a significant contribution to the work reported, whether that is in the conception, study design, execution, acquisition of data, analysis and interpretation, or in all these areas; took part in drafting, revising or critically reviewing the article; gave final approval of the version to be published; have agreed on the journal to which the article has been submitted; and agree to be accountable for all aspects of the work.

Disclosure

The authors report no conflicts of interest in this work.

References

1. Erdem I, Saritas F, Karaali R, et al. A rare cause of fever in an adult: a case of familial Mediterranean fever. *Interl Med Case Rep J*. 2018;11:37–40. doi:10.2147/IMCRJ.S148205
2. Sönmez HE, Batu ED, Özen S. Familial Mediterranean fever: current perspectives. *J Inflamm Res*. 2016;9:13–20. doi:10.2147/JIR.S91352
3. Tezcan ME, Avci M, Mercan R, Aliustaoglu M, Sargin M. MEFV gene testing may guide physicians for early diagnosis of familial Mediterranean fever. *Inter J Rheum Dis*. 2018;21(7):1452–1457. doi:10.1111/1756-185X.13258
4. Tunca M, Ozdogan H, Kasapcopur O, et al. Familial Mediterranean fever (FMF) in Turkey: results of a nationwide multicenter study. *Medicine*. 2005;84(1):1–11.
5. Ait-Idir D, Djerdjouri B. Differential mutational profiles of familial Mediterranean fever in North Africa. *Ann Human Genetics*. 2020;84(6):423–430. doi:10.1111/ahg.12404
6. Koshy R, Sivadas A, Scaria V. Genetic epidemiology of familial Mediterranean fever through integrative analysis of whole genome and exome sequences from Middle East and North Africa. *Clin Genet*. 2018;93(1):92–102. doi:10.1111/cge.13070
7. Kyejo W, Machibya M, Hooda F, et al. A private hospital's approach to treating acute familial Mediterranean fever in Dar es Salaam. *SAGE Open Med Case Rep*. 2023;11:2050313X231195964. doi:10.1177/2050313X231195964
8. Chae JJ, Wood G, Masters SL, et al. The B30. 2 domain of pyrin, the familial Mediterranean fever protein, interacts directly with caspase-1 to modulate IL-1β production. *Proc Natl Acad Sci*. 2006;103(26):9982–9987. doi:10.1073/pnas.0602081103

9. Zadeh N, Getzug T, Grody WW. Diagnosis and management of familial Mediterranean fever: integrating medical genetics in a dedicated interdisciplinary clinic. *Genet Med*. 2011;13(3):263–269. doi:10.1097/GIM.0b013e31820e27b1
10. Iuhas A, Marinău C, Niulaş L, et al. Familial Mediterranean fever in Romania: a case report and literature review. *Front Pedia*. 2025;12:1546387. doi:10.3389/fped.2024.1546387