

## A Rare Presentation of Familial Mediterranean fever; Acute Scrotum and Hydrocele Amyloidosis

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### Abstract

**Background:** Familial Mediterranean Fever (FMF) is a genetic disease characterized by recurrent febrile attacks and inflammation of serous membranes. Amyloidosis is frequent in untreated FMF patients and is also the most important complication of FMF. It is generally seen with renal, hepatic, gastrointestinal, spleen, testicular and thyroidal involvement.

**Case Presentation:** Herein, we report a case with acute scrotum and hydrocele amyloidosis as a presenting finding in a child with FMF.

**Conclusion:** Although the acute scrotum and scrotal swelling are not characteristic clinical features of FMF, this genetic disease should not be forgotten in the differential diagnosis of acute scrotum in patients of Mediterranean origins.

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**Key Words:** Amyloidosis; Scrotum; Familial Mediterranean fever; Testicular Hydrocele

### Introduction

Familial Mediterranean Fever (FMF) is an autosomal recessive inherited disorder characterized by recurrent episodes and self limited attacks of fever, peritonitis, pleuritis, arthritis and erysipelas-like erythema<sup>[1]</sup>. The most important complication of FMF is amyloidosis and is characterized by the generalized or localized extracellular deposition

of amyloid, a proteinaceous fibrillar material, in different tissues and organs. Also, amyloidosis determines the prognosis<sup>[2]</sup>. Orchitis and testicular amyloidosis are rarely reported due to FMF but can also occur with testicular amyloidosis typically being asymptomatic<sup>[3]</sup>.

Here we present a case with acute scrotum and hydrocele amyloidosis as a presenting sign of FMF.

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## Case Presentation

A previously healthy 14-year-old Turkish boy was admitted to the emergency room with swollen, painful and erythematous scrotum. At admission, he had no fever or abdominal pain.

On physical examination, translumination was positive on the scrotum. Hepatosplenomegaly was palpated. Laboratory examinations were as follow: white blood cell 19200/mm<sup>3</sup>, erythrocyte sedimentation rate 48 mm/h (normal range: 0-15) and C-reactive protein 43 mg/L (normal range: 0-5). Serum amyloid A level was 315 ng/ml (normal range: <10 ng/ml).

Scrotal and abdominal ultrasonographs showed epididymal enlargement, thickened scrotal skin, bilateral hydrocele and splenomegaly. His medical history was unremarkable for FMF. Before this attack, he had two abdominal pain attacks without fever. The abdominal pain was diagnosed as a urinary tract infection and was treated with antibiotics. He was operated to repair hydrocele. The histopathological examination of the removed hydrocele sac revealed amyloid deposition in the vessel walls. We diagnosed FMF according to Tel Hashomer criteria<sup>[4]</sup>. Genetic mutation analysis showed homozygous M694V alleles on MEFV gene. Colchicine was given for prevention and treatment of amyloidosis. Pain and swelling of scrotum resolved on day five after admission. We have been following him for two and half years. Serum amyloid A level and urine analysis were normal. He had no attacks since starting the colchicine treatment.

## Discussion

FMF can be seen at every part of the body, is characterized by fever, arthritis, abdominal and chest pain. Patients' clinical features are peritonitis (93.7%), fever (92.5%), arthritis (47.4%), pleuritis (31.2%), myalgia (39.6%) and erysipelas-like erythema (20.9%)<sup>[5]</sup>. The symptoms of FMF are related to the inflammation of serosal membranes<sup>[1]</sup>. An

extension of peritoneal sac, tunica vaginalis, could be affected by FMF at the same time. It is concluded that acute scrotum is one type of FMF attacks. There are few studies reporting scrotal swelling frequency in 4-8% of FMF patients<sup>[6,7,8]</sup>.

Only one case reported acute scrotum in a Turkish child with FMF<sup>[9]</sup>. Many other articles on FMF from Turkey did not mention acute scrotum or scrotal swelling<sup>[5]</sup>. Scrotal swelling could be a presenting sign of FMF<sup>[6]</sup>. Scrotal attack is considered to be unilateral and self limited within 24 to 72 hours. A complete recovery is expected without significant consequence<sup>[6,9]</sup>.

A recent study from Turkey indicated that FMF is the most common cause of amyloidosis in Turkish patients<sup>[2]</sup> and the highest frequency of FMF (1/123) is reported from Tokat region in Turkey<sup>[10]</sup>. Abdominal pain can be very dull and yet is sometimes very serious and seems to be an acute abdomen. It is easily misdiagnosed as appendicitis which leads to an unnecessary appendectomy<sup>[11]</sup>. Patients with typical phenotype and genetically confirmed mutations of FMF are defined as phenotype I, but phenotype II patients develop amyloidosis without any previous attacks typical of FMF<sup>[12]</sup>.

In childhood, fever or abdominal pain could be attributed to infectious disease rather than FMF so diagnosis of FMF can be postponed to older ages. Although our patient's first two abdominal pain attacks were not clear and characteristic, it can still be attributed to FMF. These reports and findings were recognized as a clue for investigating FMF in this patient. Other differential diagnosis of acute scrotum and scrotal swelling were ruled out by physical examination, laboratory and radiological test findings.

Although acute scrotum has been recognized as a feature of FMF in Jewish and Arab FMF patients, it can also be a clinical feature in Turkish FMF patients. Early diagnosis and colchicine prophylaxis are essential to prevent amyloidosis, which is the most significant, unique and lethal complication of FMF. Around the age of 40, amyloidosis is seen in 90% of FMF patients who are not treated. These patients undergo unnecessary interventions and recurrences, which lead to risk of ischemic testicular necrosis<sup>[6,13,14]</sup>.

## Conclusion

Although the acute scrotum and scrotal swelling are not characteristic clinical features of FMF, this genetic disease should not be forgotten in the differential diagnosis of acute scrotum in patients of Mediterranean origin.

## References

1. Acar BC, Yalcinkaya F, Ekim M. The pathogenesis of familial Mediterranean fever. *Turkiye Klinikleri J Pediatr*. 2006;15(4):151-5. (In Turkish)
2. Ensari C, Ensari A, Tumer N, Ertug E. Clinicopathological and epidemiological analysis of amyloidosis in Turkish patients. *Nephrol Dial Transplant*. 2005;20(8):1721-5.
3. Haimov-Kochman R, Prus D, Ben-Chetrit E. Azoospermia due to testicular amyloidosis in a patient with familial Mediterranean fever. *Hum Reprod*. 2001;16(6):1218-20.
4. Livneh A, Langevitz P, Zemer D, et al. Criteria for the diagnosis of familial Mediterranean fever. *Arthritis Rheum*. 1997;40(10):1879-85.
5. Tunca M, Akar S, Onen F, et al. Turkish FMF Study Group. Familial Mediterranean fever (FMF) in Turkey: results of a nationwide multicenter study. *Medicine (Baltimore)*. 2005; 84(1):1-11.
6. Majeed HA, Ghandour, Shahin HM. The acute scrotum in Arab children with familial Mediterranean fever. *Pediatr Surg Int*. 2000; 16(1-2):72-4.
7. Gedalia A, Mordehai J, Mares AJ. Acute scrotal involvement in children with familial Mediterranean fever. *Am J Dis Child*. 1992; 146(12):1419-20.
8. Al-Wahadneh AM, Dahabreh MM. Familial Mediterranean fever in children: a single centre experience in Jordan. *East Mediterr Health J*. 2006;12(6):818-23.
9. Makay BB, Kefi A, Unsal E. Familial Mediterranean fever in the differential diagnosis of pediatric acute scrotum. *Ege J Med*. 2007;46(2):101-3.
10. Kisacik B, Yildirim B, Tasliyurt T, et al. Increased frequency of familial Mediterranean fever in northern Turkey: a population-based study. *Rheumatol Int*. 2009;29(11):1307-9.
11. Onen F. Familial Mediterranean fever. *Rheumatol Int*. 2006;26(6):489-96.
12. Melikoğlu M, Ozdoğan H, Korkmaz C, et al. A survey of phenotype II in familial Mediterranean fever. *Ann Rheum Dis*. 2000; 59(11):910-3.
13. Pras M, Bronshpigel N, Zemer D, et al. Variable incidence of amyloidosis in familial Mediterranean fever among different ethnic groups. *Johns Hopkins Med J*. 1982;150(1):22-6.
14. Livneh A, Madgar I, Langevitz P, et al. Recurrent episodes of acute scrotum with ischemic testicular necrosis in a patient with familial Mediterranean fever. *J Urol*. 1994; 151(2):431-2.