

A Young Girl With Familial Mediterranean Fever and Abdominal Pain

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Case Report

A 9-year-old girl presented to the emergency department with an acute onset of abdominal pain and intestinal cramps. The girl's medical history was significant for a recurrent peritonitis due to familial Mediterranean fever (FMF), which had been diagnosed 1 year prior to the current illness. Notably, after prophylactic treatment with colchicine was introduced for the earlier incidents of FMF, no further attacks of abdominal pain occurred, or at least not immediately. She did not have fever, diarrhea, nausea, or other clinical signs of infections. Physical examination revealed mild unspecific abdominal tenderness with normal bowel sounds.

In the emergency department, the girl received acetaminophen for mild unexplained abdominal pain. Her symptoms ceased within a couple of minutes and she was dismissed shortly thereafter. However, within a few hours, her pain reappeared and she was admitted to inpatient care. Physical examination at that time showed diffuse abdominal tenderness to palpation along with remarkably hypoactive bowel sounds. Her blood count, clinical chemistry (electrolytes, creatinine, alanine aminotransferase) and inflammatory parameters (C-reactive protein, erythrocyte sedimentation rate) were within normal limits.

On this basis, we suspected an episode of peritonitis due to FMF and treated the girl with nonsteroidal anti-inflammatory drugs and intravenous fluids. But in contrast to previous admissions, the treatment did not alleviate her pain and she also developed no fever. Therefore, we ordered an ultrasound and computed tomography scans of the abdomen. The ultrasound revealed a widened superior mesenteric vein and widened and convoluted small mesenteric veins next to the mostly unfilled small intestine (see Figure 1). Computed tomography scans of the abdomen demonstrated the so-called whirlpool sign (see Figure 2).

Final Diagnosis

Volvulus of the small intestine.

Hospital Course

An immediate laparotomy was performed, which confirmed the volvulus due to congenital malrotation of the small intestine. Adhesiolysis and detorsion of the intestine were sufficient to restore adequate perfusion of the affected gut. Postoperative treatment and feeding was without complications. We have followed-up with the girl for 3 years postsurgery, and she has yet not had another episode of abdominal pain. The treatment with colchicine was continued without any side effects.

Discussion

FMF is an autosomal-recessive inherited auto-inflammatory disease leading to periodical attacks of fever and serosal inflammation. Most patients experience their first attack of FMF in childhood.¹ Peritonitis is the most characteristic manifestation of FMF, frequently mimicking clinical signs of an acute abdomen or ileus.¹ The peritonitis is typically accompanied by a leukocytosis and an elevation of inflammatory parameters.

Although criteria for radiologic features of FMF peritonitis (especially criteria for computed tomography) have been proposed, it is not surprising that several studies have reported on frequent surgical interventions in FMF patients.^{2,3} Appendicitis and intussusception seem to be the most important differential diagnoses in pediatric FMF patients presenting with an acute abdomen. Ultrasound has been proven to be highly valuable detecting intussusception and alternative diagnoses in a recent pediatric series.⁴

By contrast, repetitive peritoneal inflammation in FMF may result in adhesive small bowel obstruction, which has been the main gastroenterological complication in large

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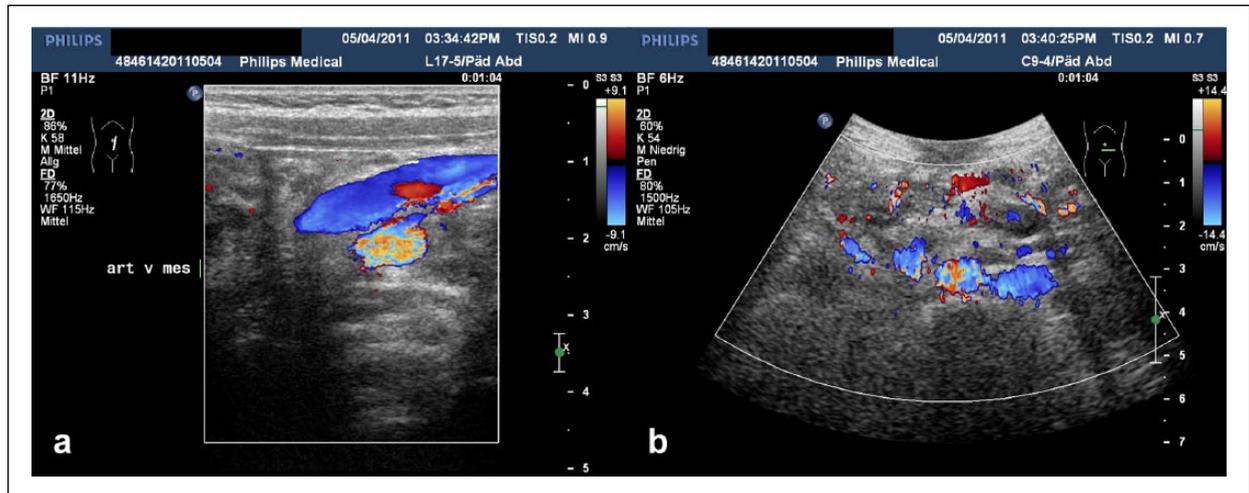


Figure 1. Initial ultrasonography of the abdomen.

Sagittal (A) and transverse ultrasonography (B) of the middle abdomen (B-mode with color Doppler) demonstrates widened superior mesenteric vein (●) (A) and widened and convoluted mesenteric veins (■) next to mostly unfilled small intestine (B).

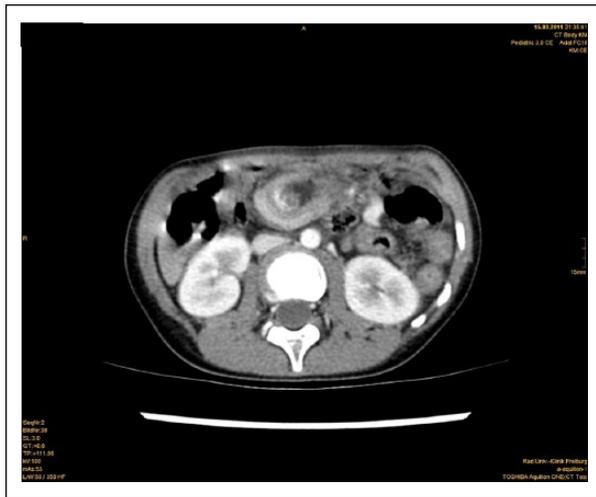


Figure 2. Computed tomography of the abdomen.

Transverse computed tomography image of the abdomen (contrast enhanced) reveals intestinal volvulus with complete twisting of a loop of intestine around its mesenteric attachment site (so-called “whirlpool” sign).

pediatric series.^{5,6} The incidence of adhesive small bowel obstruction in children with FMF has been reported as 3%.⁶

Unfortunately though, there are no international guidelines regarding when to initiate additional diagnostic tests in order to exclude other entities causing acute abdominal pain in FMF patients.

Symptomatic malrotation is a rare congenital disorder affecting ~1 in 6000 births. Associated malformations of the gastrointestinal tract are detectable in 30% to 60% of the affected children.⁷ A malrotation usually

presents in early infancy with periodical signs of intestinal obstruction, like abdominal distension, pain, and vomiting. Volvulus is the most common complication of malrotation, affecting ~20% of patients (especially neonates).⁷ It might present as acute abdomen. Nevertheless, the risk of developing volvulus in patients with a congenital malrotation has not been determined so far.

Upper gastrointestinal series remain the gold standard in detection of malrotation, although diagnostic criteria for ultrasound (ie, abnormal position of the superior mesenteric vein, dilated mesenteric vessels twisting around the base of the mesenteric pedicle, “whirlpool” sign) have been proposed.⁸ Notably, an upper gastrointestinal series would not have been sufficient to detect volvulus in our patient.⁹

In most patients, early operative intervention is the only therapy that would lead to an effective resolution of symptoms of malrotation and prevention of volvulus.⁷ When diagnosis of volvulus can be established, early surgery prevents substantial injury to the affected gut.⁹

Conclusion

Unusual presentations of well-defined diseases as well as the absence of pathognomonic features (such as fever and elevated inflammatory parameters in FMF) should alert physicians to the possibility of pathologies other than expected. Additional diagnostic tests should be requested to exclude harmful illnesses.

Surgical exploration of an acute abdomen should not be delayed in FMF patients, even if abdominal symptoms are compatible with underlying FMF disease. Volvulus due to congenital malrotation is one

differential diagnosis of abdominal pain in FMF patients, which, to the best of our knowledge, has not been reported previously.

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Declaration of Conflicting Interests

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