

## Coexistence of Familial Mediterranean Fever and Acute Intermittent Porphyria

Gebe Bir Hastada Ailesel Akdeniz Ateşi ve Akut Intermittan Porfiri Birlikteliği

Faruk AKTÜRK, Yaşar ÇALIŞKAN, Bahar ESEN

*Department of Internal Medicine, Medical Faculty of İstanbul University, İstanbul, Turkey*

A 19-year-old woman in her sixth month of pregnancy was referred to our clinic with stabbing chest pain, abdominal pain, fatigue, fever, and joint pain. She was pale, and distressed, and her body temperature was 38.5 °C. Her lungs were clear to auscultation. Her pulse rate was 90/min, her blood pressure was 100/60 mmHg, and a midsystolic murmur was heard in all cardiac areas.

Laboratory investigation indicated a total leukocyte count of 8500/mm<sup>3</sup>, a hemoglobin level of 8.5 g/dl, a platelet count of 400.000/mm<sup>3</sup>, an erythrocyte sedimentation rate (ESR) of 120 mm/h, a C-reactive protein (CRP) level of 30 mg/l, and mild hyponatremia (132 mEq/l). In addition, her serum D-dimer level was up to three times higher than normal. The patient underwent thoracic and abdominal magnetic resonance imaging (MRI) as well as thyroid function tests and transthoracic echocardiography (TTE), and all yielded normal results. Her blood and urine cultures were sterile.

A detailed history, taking into special consideration inflammatory rheumatic diseases, revealed recurrent attacks of ankle and knee arthritis as well as an erythematous skin rash during childhood. Antinuclear antibody (ANA) and anti neutrophil cytoplasmic antibody (ANCA) tests were negative. Mediterranean fever (MEFV) gene

analysis showed a homozygous M694V mutation that indicated familial Mediterranean fever (FMF). After beginning treatment with 1.5 mg/day colchicine, vomiting and watery diarrhea occurred related to the medication. Intravenous fluids, metoclopramide, and ondansetron were then given for severe nausea and vomiting.

The patient developed severe depression with homicidal and suicidal thoughts. She later suffered from agitation and seizures, and haloperidol was added to her therapy. A change in the color of her urine to a dark reddish appearance was noticed. Porphobilinogen was found to be strongly positive in the analysis of a spot urine sample, and her uroporphyrin, corpoporphyrin and total porphyrin levels were found to be elevated in a 24-hour urine collection. Taking all of these factors into account, a concomitant diagnosis of acute intermittent porphyria (AIP) was indicated. All medications that might trigger porphyria attacks were stopped, and intravenous 300 g/day dextrose was given. Her symptoms resolved after a few days, and at term, she gave birth to a healthy baby. Since the delivery, she has been free of symptoms.

Pregnancy may affect the normal course of FMF with some women enjoying an attack-free period while others may experience devastating attacks with

**Received:** March 14, 2012 **Accepted:** April 10, 2012

**Correspondence:** Faruk Aktürk, M.D. İstanbul Üniversitesi İstanbul Tıp Fakültesi İç Hastalıkları Anabilim Dalı, 34093 Fatih, Çapa, İstanbul, Turkey.

Tel: +90 216 - 545 33 84 e-mail: farukakturk@gmail.com

©2012 Turkish League Against Rheumatism. All rights reserved.

high frequency. When this occurs, there is the risk of peritonitis which could lead to an eventual abortion.<sup>[1,2]</sup>

Acute intermittent porphyria may cause periodic abdominal pain, and it is one of the hepatic porphyrias in which characteristically no photosensitivity is seen. This is due to mutations in the gene encoding for uroporphyrinogen I synthetase.<sup>[3,4]</sup> In our case, diagnosis was made after the recognition of dark urine and higher than normal urine porphobilinogen, corpoporphyrin and total porphyrin levels. Abdominal pain, peripheral neuropathy, and changes in mental status are the classic triad featured in an acute attack. Environmental factors, such as prescribed or illegal drugs, a low-calorie diet, dehydration, infection, and variations in hormone levels, may also precipitate acute attacks. In women, relapses are more likely before menstruation and during pregnancy.<sup>[5]</sup>

Acute AIP attacks are treated with intravenous glucose (300-500 g/day). Intravenous hematin or heme arginate are also used in severe cases. Our patient dramatically responded to a glucose infusion and the discontinuation of drugs that were triggering her porphyria attacks.

### Declaration of conflicting interests

The authors declared no conflicts of interest with respect to the authorship and/or publication of this article.

### Funding

The authors received no financial support for the research and/or authorship of this article.

---

## REFERENCES

---

1. Ben-Chetrit E, Ben-Chetrit A, Berkun Y, Ben-Chetrit E. Pregnancy outcomes in women with familial Mediterranean fever receiving colchicine: is amniocentesis justified? *Arthritis Care Res (Hoboken)* 2010;62:143-8.
2. Ofir D, Levy A, Wiznitzer A, Mazor M, Sheiner E. Familial Mediterranean fever during pregnancy: an independent risk factor for preterm delivery. *Eur J Obstet Gynecol Reprod Biol* 2008;141:115-8.
3. Kauppinen R, Mustajoki P. Prognosis of acute porphyria: occurrence of acute attacks, precipitating factors, and associated diseases. *Medicine (Baltimore)* 1992;71:1-13.
4. Kushner JP. Laboratory diagnosis of the porphyrias. *N Engl J Med* 1991;324:1432-4.
5. Farfaras A, Zagouri F, Zografos G, Kostopoulou A, Sergentanis TN, Antoniou S. Acute intermittent porphyria in pregnancy: a common misdiagnosis. *Clin Exp Obstet Gynecol* 2010;37:256-60.