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## Mobile right heart thrombus as a manifestation of homozygous mutation of MTHFR 1298 A > C



*Homozigot MTHFR 1298 A > C mutasyonunun  
belirtisi olarak mobil sağ atriyal trombüs vakası*

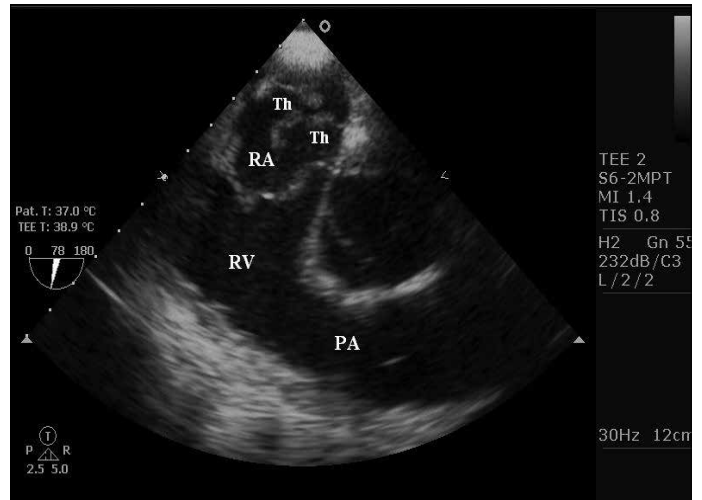
Dear Editor,

Mobile right heart thrombus (MRHT) is uncommon pathology but the true prevalence is still unknown. Previous studies reported that MRHT occurs in 7% to 18% of patients with pulmonary embolism with high mortality rate (44.7%) (1). The main manifestations of venous thromboembolism (VTE) are deep venous thrombosis (DVT) and pulmonary embolism. In addition, genetic factors play an important role in pathogenesis of VTE. The relationship between common genetic mutations such as factor V Leiden, prothrombin factor II G 20210A, methylenetetrahydrofolate reductase (MTHFR), deficiencies of protein C, protein S, and antithrombin III, and VTE have been reported (2).

A 34-year-old man admitted to the emergency department because of sudden onset of dyspnea. He had no previous history of both VTE and acute coronary syndrome. He denied any trauma, history of malignancy, recent surgery, and any drug usage. Only 10 days before, he had fracture of toe, which did not require plaster cast and immobilization. Admission physical examination was unremarkable. Baseline 12-lead electrocardiogram (ECG) revealed sinus rhythm and S1Q3T3 sign. Duplex scan of the lower extremities was also normal. Because of suspicion of pulmonary embolism, bedside transthoracic echocardiography (TTE) was performed, which revealed mobile right atrial mass. Left ventricular ejection fraction was normal (60%). Right ventricle was not enlarged and estimated systolic pulmonary arterial pressure was 28 mmHg. Because of poor imaging quality with TTE, transesophageal echocardiography (TEE) was performed and two hypermobile and snake-like thrombi in the right atrium were demonstrated (Fig. 1 and Video 1. See corresponding video/movie images at [www.anakarder.com](http://www.anakarder.com)). Laboratory parameters were within normal limits. Protein C and S levels were also normal. Homocysteine was slightly elevated: 19.6 µmol/L (5.5-14 µmol/L). Upon genetic testing, there were no mutations in the factor V Leiden (G1691A), factor II (G20210A), and MTHFR (C677T). Only homozygous mutation of MTHFR (A1298C) was detected. Pulmonary computed tomography angiography revealed bilateral lower lobe pul-

monary embolism (Fig. 2). Because of hemodynamic stability and no evidence of RV strain, anticoagulation with heparin was started. After 10 days of hospitalization, control TEE was performed and right atrial thrombi were markedly decreased (Fig. 3 and Video 2. See corresponding video/movie images at [www.anakarder.com](http://www.anakarder.com)). The patient was discharged with warfarin treatment for indefinite time.

The relationship between both C677T and A1298C polymorphisms of the MTHFR gene and VTE have been analyzed before (3). However, there are conflicting results regarding the role of MTHFR gene mutation in VTE pathogenesis (4). In cases of homozygous MTHFR mutations, hyperhomocysteinemia may occur. The relationship between hyperhomocysteinemia and VTE is still unknown and conflicting results are present. Auerbach et al. (5) have reported that homozygous MTHFR mutation that leads to hyperhomocysteinemia can increase the risk of VTE up to 2.5-folds. Therefore, in the present case, we postulated that the possible cause of right atrial thrombi is MTHFR A1298C homozygous mutation because no other predisposing factors were present in our patient. In addition, previous cases of VTE were presented with heterozygous MTHFR mutation. However, our patient had homozygous MTHFR mutation and, right heart thrombus as a manifestation of homozygous

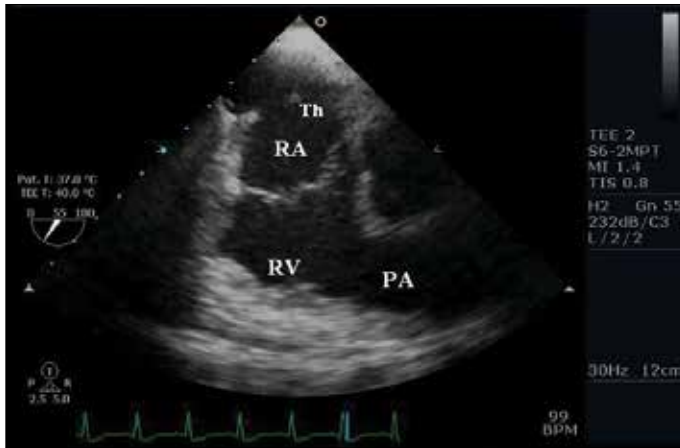


**Figure 1. Pretreatment transesophageal echocardiography showing two mobile thrombi in the right atrium**

PA - pulmonary artery, RA - right atrium, RV - right ventricle, Th - thrombus



**Figure 2. Pulmonary computed tomography showing bilateral thrombi in the lower lobe**



**Figure 3. Post-anticoagulation therapy, transesophageal echocardiography showing markedly reduced thrombi in the right atrium**

PA - pulmonary artery, RA - right atrium, RV - right ventricle, Th - thrombus

MTHFR mutation has not been reported before. In conclusion, we suggest analyzing genetic mutations in patients with VTE who had not any predisposing factors. Because diagnosis of genetic mutation associated with VTE will require long-term anticoagulation.

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**Video 1. Pretreatment transesophageal echocardiography showing two mobile thrombi in the right atrium**

PA - pulmonary artery, RA - right atrium, RV - right ventricle, Th - thrombus

**Video 2. Post-anticoagulation therapy, transesophageal echocardiography showing markedly reduced thrombi in the right atrium**

PA - pulmonary artery, RA - right atrium, RV - right ventricle, Th - thrombus

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## Paroxysmal supraventricular arrhythmias during hypokalemic episodes in a patient with hypokalemic periodic paralysis

*Hipokalemik periyodik paralizili bir hastada hipokalemik epizodlar sırasında gelişen paroksizmal supraventriküler aritmiler*

Dear Editor,

A 21-year-old female patient was admitted to our hospital with severe muscle weakness, fatigue, unable to move all extremities and palpitation following a high carbohydrate meal. The patient described similar symptoms a week ago, which she recovered spontaneously in 48 hours. Her past and family history was unremarkable. Physical examination was notable for flaccid tetraparesis, decreased deep tendon reflexes, with sparing of the facial, oropharyngeal and respiratory muscles. Sensory testing was intact. Thyroid and other system examinations were unremarkable. Electrocardiography (ECG) on admission revealed supraventricular tachycardia (180 bpm) (Fig. 1A). Initial laboratory tests showed a potassium level of 2.67 mEq/L (normal range 3.5-5.1 mEq/L); all the other routine examinations and thyroid hormone levels were normal. She presented sinus rhythm after intravenous potassium replacement and diltiazem (12.5 mg). Control potassium level showed 3.78 mEq/L. Electrophysiological study revealed dual AV nodal physiology, inability to induce any tachycardia and no ablation therapy. She discharged uneventfully. While she was asymptomatic for 2 months, the patient admitted to emergency room with palpitation again. ECG on admission showed atrial tachycardia (166 bpm) (Fig. 1B). In addition, biochemistry tests showed potassium level of 2.78 mEq/L. Her palpitation was resolved after intravenous potassium replacement again. She was referred for investigation of the reasons of hypokalemia. Serum potassium levels were normal in between emergency admissions. Also serum magnesium, sodium, calcium levels and thyroid function tests were in normal limits. Urinary potassium level was decreased (24 mEq/L). Urinary potassium/creatinine ratio was 0.50. Transtubular potassium gradient was 6.8 (normal range: 7-9). Arterial blood gas analysis showed no metabolic alkalosis. Serum renin, aldosterone and ACTH levels were normal. Adrenal gland imaging with computed tomography revealed normal findings. So, hypokalemic periodic paralysis was considered in differential diagnosis, which was confirmed by genetic testing (mutation in SCN4A, Arg669H). The patient was discharged with oral potassium supplement (potassium citrate 2.17 gr/day plus potassium carbonate 2.0 gr/day) and avoidance of strenuous exercise and high carbohydrate diet. The 6-months follow-up was free of new paralysis and palpitation episodes.

Hypokalemic periodic paralysis is an autosomal dominant disorder which is accompanied by muscle weakness/paralysis and hypokalemia. Attacks can be induced by exercise, carbohydrate-rich meals



**Figure 1. Initial electrocardiography showing supraventricular tachycardia (180 bpm) on first admission (A). Atrial tachycardia (166 bpm) was also seen on second admission (B)**