Aortic Thrombus and Acute Pulmonary Embolism in an Individual Heterozygous for the MTHFR C677-T Mutation

A 57-year-old man, without cardiovascular risk factors, was admitted to our Intensive Coronary Unit for dyspnea. He had a brother in warfarin therapy for deep venous thrombosis and his daughter, with a history of several miscarriages, had a cytosine-to-thymidine substitution, at nucleotide 677 (C677T) of the 5,10-methylenetetrahydrofolate reductase (MTHFR) gene.

On admission, he had slightly elevated blood pressure (140/100 mm Hg). The ECG showed sinus tachycardia (114/min) but was otherwise normal. Laboratory abnormal findings were: troponin I, 0.44 ng/mL; D-dimer, 1733 ng/L, and pO₂=66.8 mm Hg. Thoracic CT scan (Figure 1 upper panel) and transesophageal echocardiography (Figure 1 lower panel) showed pulmonary emboli and a thrombus (10×18 mm) of aortic isthmus, which led to the diagnosis of acute pulmonary embolism and aortic thrombus.

The same gene mutation (C677-T MTHFR) was detected by polymerase chain reaction.

Transesophageal echocardiogram after 40 days of warfarin therapy documented the disappearance of aortic thrombus (Figure 2).

Recent studies showed that C677T gene polymorphism is associated with an increased risk of arterial disease and a major risk of pulmonary embolism.