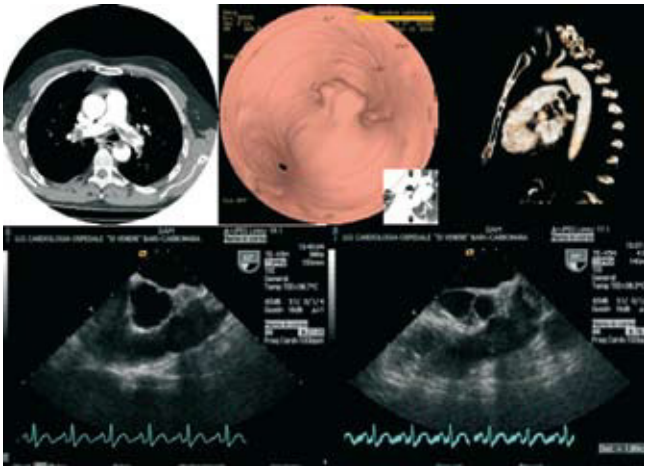

Figure 1.

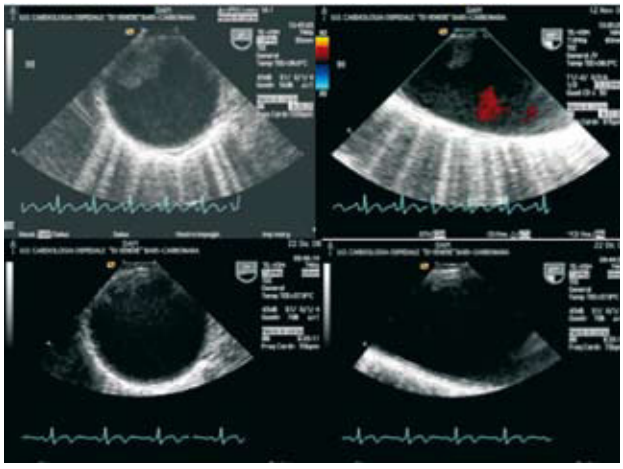

Figure 2.

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 pO_2=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 polimophism is associated with an increased risk of arterial disease and a major risk of pulmonary embolism.

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