Abstract

Background
Acute myocardial infarction (AMI) in young women represent an extreme phenotype associated with a higher mortality compared with similarly aged men. Prothrombotic gene variants could play a role as risk factors for AMI at young age.

Methods
We studied Factor V Leiden, FII G20210A, MTHFR C677T and beta-fibrinogen -455G>A variants by real-time PCR in 955 young AMI (362 females) and in 698 AMI (245 females) patients. The data were compared to those obtained in 909 unrelated subjects (458 females) from the general population of the same geographical area (southern Italy).

Results
In young AMI females, the allelic frequency of either FV Leiden and of FII G20210A was significantly higher versus the general population (O.R.: 3.67 for FV Leiden and O.R.: 3.84 for FII G20210A; $p<0.001$). Among AMI patients we showed only in males that the allelic frequency of the MTHFR C677T variant was significantly higher as compared to the general population. Such difference was due to a significantly higher frequency in AMI males of the MTHFR C677T variant homozygous genotype (O.R. 3.05).

Discussion and conclusion
Our data confirm that young AMI in females is a peculiar phenotype with specific risk factors as the increased plasma procoagulant activity of FV and FII. On the contrary, the homozygous state for the 677T MTHFR variant may cause increased levels of homocysteine and/or an altered folate status and thus an increased risk for AMI, particularly in males. The knowledge of such risk factors (that may be easily identified by molecular analysis) may help to improve prevention strategies for acute coronary diseases in specific risk-group subjects.

Keywords:
Young AMI; Gender; AMI; Gene variants; Mutations; Prothrombotic variants; Genetic predisposition