
ABSTRACT. We investigated mutations and polymorphisms of the coronary artery disease (CAD)-related myocyte enhancer factor 2A (MEF2A) gene in a Chinese population. Polymerase chain reaction-single-strand conformation polymorphism and DNA sequencing were used to detect exon 11 of the MEF2A gene in 210 Hubei patients with CAD and 190 healthy controls. The following mutations were identified: a) a synonymous heterozygous mutation (147191G→T) combined with a 6-base deletion (147123-147128); b) a synonymous heterozygous or homozygous mutation (147191G→T) combined with a 9-base deletion (147123-147131); c) a synonymous mutation (147143G→A); d) a synonymous mutation (147191G→T) combined with an 18-base deletion (147111-147128); and e) a 21-base deletion (147108-147128). Mutations (a) and (b) and a 3-base deletion (147126-147128) with
or without the synonymous heterozygous mutation (147191G→T) occurred in more than 1% of controls. However, mutations (c), (d), and (e) were not observed in the control group. The polymorphism in exon 11 of the MEF2A gene was observed in the Chinese population. Six or seven amino acid deletions and synonymous mutations (147143G→A) may be correlated with susceptibility to CAD.

**Key words:** Coronary artery disease; MEF2A; Gene mutation; Polymorphism