conferenceseries.com

J Cardiovasc Dis Diagn 2018, Volume 6
DOI: 10.4172/2329-9517-C1-003

Factor XII gene mutations in acute myocardial infarction patient with Factor XII defect

Fan Liu, Bing Xiao and Xiuchun Yang
Hebei Medical University, China

Statement of the Problem: A 51-year-old man had a myocardial infarction. The patient disclosed medical history of hypertension and coagulation Factor XII defect. Later coronary angiography showed triple vessels lesions. In the present study, the risk of coronary artery diseases related to Factor XII gene was investigated.

Method: Factor κ coagulation activity was detected by clotting test and 46C>T polymorphism was genotyped using a Restriction Fragment Length Polymorphism (RFLP) method. Gene mutations were analyzed in patients with DNA sequencing. Expression plasmids were constructed by site-directed mutagenesis based on the wild-type and transiently transfected into 293T cells. Factor κ activity of the expression products were tested in the cell lysates.

Findings: A functional promoter polymorphism F12 46C/T (rs1801020) that results in decreased translation efficiency was investigated and a homozygote TT was revealed. Genetic analysis of Factor XII polymorphisms displayed that a non-synonymous mutation p.D562G and a stop coden p.W258X were disclosed in this patient with myocardial infarction. The results of the transfection revealed that Factor antigens in cell lysates of mutant protein W258X was significantly lower.

Conclusion: Rather than bleeding tendency, Factor XII defect may be associated with possible thrombotic disorders. Both p.D562G and p.W258X mutations may contribute to the pathogenesis of acute myocardial infarction patient with Factor XII defect.

liufantrail@163.com

Notes: