Deteksi polimorfisme loss of function R46l pada gen proprotein convertase subtilisin-kexin type-9 dan hubungannya dengan luaran kardioserebrovaskular pada pasien infark miokard akut disertai elevasi segmen ST yang menjalani intervensi koroner perkutan pri = Detection of proprotein convertase subtilisin kexin type-9 gene R46L loss of function polymorphism and its association with major adverse cardiocerebrovascular outcomes in acute ST-segment elevation myocardial infarction undergoing primary percutaneous co

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Abstrak

ABSTRAK
Latar belakang: PCSK9 merupakan protein yang berperan dalam regulasi kadar kolesterol LDL darah. PCSK9 diketahui memiliki mekanisme kerja lain yang melibatkan proses inflamasi, peningkatan Lp(a), aktivasi jaras protrombotik dan platelet, metabolisme triglyceride-rich lipoprotein, serta modifikasi plak yang juga dapat berperan dalam patogenesis berbagai spektrum penyakit aterosklerotik, termasuk IMA-EST. Kemajuan dalam strategi penatalaksanaan IMA-EST telah berhasil meningkatkan kesintasan. Polimorfisme R46L gen PCSK9 diketahui memiliki efek proteksi terhadap risiko kardiovaskular. Pada pasien infark miokard, prevalensi pembawa karier mutan R46L sebesar 2,14%. Dalam observasi pasien infark miokard akut didapatkan proporsi pasien yang memiliki kesintasan yang panjang. Polimorfisme R46L gen PCSK9 dipikirkan dapat memiliki peranan dalam mempertahankan kesintasan pasien-pasien tersebut. Tujuan: Penelitian ini bertujuan untuk mempelajari hubungan antara polimorfisme R46L gen PCSK9 pada pasien IMA-EST yang menjalani IKPP dengan luaran kardioserebrovaskular mayor. Metode: Sebanyak 601 pasien dengan IMA-EST yang menjalani IKPP diperiksakan polimorfisme R46L gen PCSK9 pada saat admisi. Data luaran kardioserebrovaskular mayor dan data penunjang lain didapatkan dari rekam medik dan follow-up melalui telepon. Hasil: Tidak ditemukan varian mutan (GT dan TT) polimorfisme R46L gen PCSK9 pada pasien IMA-EST yang menjalani IKPP sehingga analisa hubungan polimorfisme R46L gen PCSK9 terhadap luaran kardioserebrovaskular mayor tidak dapat dilakukan. Kesimpulan: Pada pasien IMA-EST yang menjalani IKPP di RS Jantung Harapan Kita, tidak ditemukan varian mutan R46L gen PCSK9. Analisa hubungan polimorfisme R46L gen PCSK9 terhadap luaran kardioserebrovaskular mayor tidak dapat dilakukan.
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ABSTRACT
Background: PCSK9 is a protein molecule that regulates serum LDL cholesterol level. Recent data suggest that PCSK9 activity may also work through other mechanisms, such as inflammation, increased Lp(a), triglyceride-rich lipoprotein metabolism, activation of prothrombotic pathways and platelets, and modification of atherosclerotic plaque, which may contribute to the pathogenesis of atherosclerotic diseases, including STEMI. Advances in the management of STEMI have succeeded in increasing survival. Polymorphism R46L of PCSK9 gene has been known to have protective effect on cardiovascular risks. In patients with myocardial infarction, the prevalence of R46L mutation carriers was 2.14%. In the longterm

observation of acute coronary syndrome patients, a proportion of patients experienced longer survival. Polymorphism R46L of PCSK9 gene may play a role in longterm survival. Objective: The aim of this study is to evaluate the association between plasma polymorphism R46L of PCSK9 gene with MACCE in STEMI patients who underwent primary PCI. Methods: In total, 601 patients with STEMI who were treated with primary PCI had their plasma sample drawn during admission and evaluated for polymorphism R46L of PCSK9 gene. MACCE and other supportive data were taken from the medical records and telephone follow-up. Results: In this study, no polymorphism R46L of PCSK9 gene was detected. Therefore, its association with MACCE could not be further analysed. Conclusion: There was no polymorphism R46L of PCSK9 gene detected in STEMI patients treated with primary PCI. The analysis of its association with MACCE could not be conducted.