

Asosiasi alel risiko SNP rs290487 dan rs290481 gen transcription factor 7-Like 2 (TCF7L2) terhadap sindrom metabolik pada populasi Bali = Association of transcription factor 7-Like 2 (TCF7L2) gene SNP rs290487 and rs290481 risk alleles with metabolic syndrome in Balinese population

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Deskripsi Lengkap: <https://lib.ui.ac.id/detail?id=20493273&lokasi=lokal>

Abstrak

Sindrom metabolik merupakan sekumpulan faktor risiko yang meningkatkan peluang kemunculan penyakit kardiovaskular. Sindrom tersebut muncul akibat interaksi faktor lingkungan dan faktor genetik. Beberapa penelitian menemukan bahwa single nucleotide polymorphisms (SNPs) pada gen Transcription Factor 7-Like 2 (TCF7L2) merupakan variasi yang paling berpengaruh terhadap kemunculan sindrom metabolik. Namun, studi pada SNP rs290487 dan rs290481 belum banyak dilakukan. Penelitian ini bertujuan untuk mengetahui asosiasi alel risiko rs290487 dan rs290481 dengan sindrom metabolik pada populasi Bali. Studi ini menggunakan 565 sampel (321 sampel laki-laki dan 244 sampel perempuan) yang berasal dari empat desa di Provinsi Bali. Keberadaan SNP dideteksi menggunakan teknik amplification refractory mutation system polymerase chain reaction (ARMS PCR) dengan primer yang telah didesain khusus. Analisis asosiasi SNP rs290487 dan rs290481 dilakukan menggunakan uji regresi logistik ordinal. Hasil analisis menunjukkan MAF (alel T) SNP rs290487 dan rs290481 masing-masing sebesar 0,56 dan 0,53. Alel C pada masing-masing SNP merupakan alel risiko bagi peningkatan kadar gula darah puasa (GDP) dan kemunculan sindrom metabolik pada sampel perempuan di populasi Bali. Selain itu, analisis haplotype yang dilakukan menemukan bahwa haplotype TTT dari rs290487, rs290481, dan rs7903146, berasosiasi dengan peningkatan TG ($p = 0,011$) dan kemunculan sindrom metabolik ($p = 0,003$)

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Metabolic syndrome (MetS) is a cluster of risk factors that raises risk of cardiovascular disease. MetS can occur due to an interaction between environmental and genetic factors. Several studies have shown that single nucleotide polymorphisms (SNPs) of Transcription Factor 7-Like 2 (TCF7L2) gene are significantly associated with MetS. Nevertheless, studies on SNP rs290487 and rs290481 have not yet been explored widely. This study aimed to investigate the association of rs290487 and rs290481 risk alleles with MetS in Balinese population. A total of 565 archive samples (321 males and 244 females) from four villages in Bali province were included in this study. All subjects were genotyped using amplification refractory mutation system polymerase chain reaction (ARMS PCR) method with specifically designed primers. Association between rs290487 and rs290481 with MetS were analyzed using ordinal logistic regression test. The results showed that MAF of rs290487 and rs290481 are 0,56 and 0,53, respectively. C-allele of both SNPs were risk alleles for elevated fasting plasma glucose (FPG) level and development of MetS in Balinese women. Furthermore, haplotype TTT of rs290487, rs290481, and rs7903146, were significantly associated with elevated TG level ($p = 0,011$) and development of MetS ($p = 0,003$).