# Uji coba metode identifikasi Single Nucleotide Polymorphisms (SNP) rs434 pada gen Angiotensin Converting Enzyme (ACE) menggunakan rhAmp SNP genotyping system = Trial of Single Nucleotide Polymorphisms (SNP) rs4343 gen Angiotensin Converting Enzyme (ACE) identification method using rhAmp SNP genotyping system

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Abstrak

Dalam beberapa tahun terakhir, beberapa penelitian telah membuktikan bahwa variasi genetik berkontribusi terhadap kesehatan fisik dan mental manusia serta mempengaruhi hasil terapinya. Sejumlah penelitian juga telah menyelidiki peran Single Nucleotide Polymorphisms (SNP) dalam farmakodinamik dan farmakokinetik kemoterapi seperti fluoropyrimidine, meskipun implementasi hasil yang diperoleh masih sederhana. Pada akhir tahun 2019, WHO menemukan kasus pneumonia baru yang disebabkan oleh Severe Acute Respiratory Syndrome Coronavirus – 2 (SARS-CoV-2), yang kemudian dinyatakan sebagai pandemi coronavirus disease 2019 (COVID-19). Regulasi virus dipengaruhi oleh gen Angiotensin Converting Enzyme (ACE) dan Renin-Angiotensin System (RAS). Tujuan dari penelitian ini adalah untuk melakukan uji coba deteksi SNP menggunakan protokol real-time quantitative Polymerase Chain Reaction (qPCR) dengan rhAmp SNP genotyping system pada rs4343. SNP rs4343 terletak di ekson 17 gen ACE dengan alel G dan A. Sampel darah diperoleh dari penyintas COVID-19 dan DNA genom diekstraksi dari sampel darah. Penelitian ini menggunakan fragmen gen gBlocks™ sebagai kontrol positif dan campuran qPCR terdiri dari rhAmp Master Mix, rhAmp Reporter Mix, dan rhAmp rs4343 assay. Hasil disajikan dalam bentuk plot allelic discrimination dan memberikan hasil yang kurang memuaskan untuk deteksi rs4343. Hanya 27 sampel terdeteksi memiliki alel G homozigot dari 50 sampel yang diuji dimana 23 sampel lainnya tidak berhasil dideteksi.

.....In recent years, several studies have shown that genetic variation contributes to both the physical and mental health of humans and affects the outcome of therapy. A number of studies have also been carried out on the role of Single Nucleotide Polymorphisms (SNP) in the pharmacodynamics and pharmacokinetics of chemotherapy such as fluoropyrimidines, although the implementation of the results obtained is yet simple. At the end of 2019, WHO found a new case of pneumonia caused by Severe Acute Respiratory Syndrome Coronavirus – 2 (SARS-CoV-2), which was later declared a pandemic coronavirus disease 2019 (COVID-19). The regulation of the virus was influenced by the Angiotensin Converting Enzyme (ACE) and Renin-Angiotensin System (RAS) genes. The aim of this study was to carry out a SNP detection trial employing a protocol of a real-time quantitative Polymerase Chain Reaction (qPCR) performing rhAmp SNP genotyping assay on rs4343. SNP rs4343 is located in exon 17 of the ACE gene with alleles G and A. Blood samples were obtained from COVID-19 survivors and the genomic DNAs were extracted from them. This study used gene fragment gBlocksTM as the positive control and the qPCR mix consisted rhAmp Master Mix, rhAmp Reporter Mix, and rhAmp rs4343 assay. The results were presented in the form of allelic discrimination plots and is not satisfied to use the method to detect rs4343.There are only 27 samples were detected to have the homozygous G allele out of 50 samples where the other 23 samples were undetermined.