

Identifikasi Varian Gen *CYP11B1/CYP11B2* pada Kasus Hipertensi di Keluarga Magelang Jawa Tengah

INTISARI

Hipertensi monogenik merupakan hipertensi yang disebabkan oleh mutasi gen tunggal yang diwariskan melalui pola pewarisan Mendelian. Hipertensi monogenik menyumbang sekitar 30% kasus hipertensi secara keseluruhan. Patofisiologi yang ditimbulkan dapat berupa peningkatan absorpsi natrium yang akan mengarah pada ekspansi volume plasma darah, produksi hormon aldosteron yang terlalu banyak, dan defisiensi enzim yang bertanggung jawab dalam regulasi sintesis hormon steroid adrenal maupun represi hormon tersebut. Penelitian ini bertujuan untuk mengetahui ada tidaknya mutasi ekson 9 gen *CYP11B1* pada subjek yang diteliti, ada tidaknya gen *chimeric CYP11B1/CYP11B2* pada subjek yang diteliti dan hasil pengujian hormon pada salah satu subjek penelitian. DNA didapatkan dari saliva. Mutasi ekson 9 *CYP11B1* dideteksi dengan PCR yang kemudian disekuensing. Penyejajaran dilakukan dengan program MEGA11 dan DnaSP. Gen *chimeric CYP11B1/CYP11B2* dideteksi menggunakan metode *Two-step PCR* dengan hasil visualisasi elektroforesis sekitar 4 kbp. Analisis mutasi ekson 9 *CYP11B1* dilakukan pada 25 subjek sedangkan deteksi gen *chimeric CYP11B1/CYP11B2* dilakukan pada 23 subjek. Uji *chi-square* dilakukan untuk mengetahui hubungan ditemukannya mutan dengan kasus hipertensi pada subjek yang diteliti menggunakan *software* SPSS versi 26. Hasil penelitian menunjukkan tidak ditemukan mutasi ekson 9 *CYP11B1* pada 25 subjek. Gen *chimeric CYP11B1/CYP11B1* ditemukan pada 16 dari 23 subjek. Hasil pengujian hormon menunjukkan adanya peningkatan hormon adenokortikoid (ACTH) pada salah satu subjek sedangkan kadar aldosteron, aktivitas plasma renin dan kalium masih dalam batas normal.

Kata kunci: hipertensi monogenik, *CYP11B1/CYP11B2*, gen *chimeric*

Identification of *CYP11B1/CYP11B2* Gene Variants in Hypertension Cases in the Magelang Family, Central Java

ABSTRACT

Monogenic hypertension is hypertension caused by a single gene mutation that is inherited through a Mendelian inheritance pattern. Monogenic hypertension accounts for approximately 30% of hypertension cases overall. The resulting pathophysiology can be in the form of increased sodium absorption which will lead to expansion of blood plasma volume, excessive production of the hormone aldosterone, and deficiency of enzymes responsible for regulating the synthesis of adrenal steroid hormones and repression of these hormones. This study aims to determine whether there is a mutation in exon 9 of the *CYP11B1* gene in the subjects studied, whether there is a chimeric *CYP11B1/CYP11B2* gene in the subjects studied and the results of hormone testing in one of the research subjects. DNA is obtained from saliva. *CYP11B1* exon 9 mutations were detected by PCR and then sequenced. Alignment was carried out with the MEGA11 and DnaSP programs. The chimeric *CYP11B1/CYP11B2* gene was detected using the Two-step PCR method with electrophoresis visualization results of around 4 kbp. *CYP11B1* exon 9 mutation analysis was carried out in 25 subjects, while *CYP11B1/CYP11B2* chimeric gene detection was carried out in 23 subjects. The *chi-square* test was carried out to determine the relationship between the discovery of mutants and cases of hypertension in the subjects studied using SPSS version 26 software. The results of the study showed that no *CYP11B1* exon 9 mutations were found in 25 subjects. The chimeric *CYP11B1/CYP11B1* gene was found in 16 of 23 subjects. The results of hormone testing showed an increase in adrenocorticoid hormone (ACTH) in one of the subjects, while aldosterone levels, plasma renin activity and potassium were still normal.

Keywords: *monogenic hypertension, CYP11B1/CYP11B2, chimeric gene*