

Hubungan kadar androsteron dan etiokolanolon urin dengan kadar 17-Hidroksiprogesteron pada pasien hiperplasia adrenal kongenital defisiensi 21-Hidroksilase dalam pemantauan tata laksana = Correlation between androsterone, etiocholanolone urine with 17-hydroxyprogesterone in congenital adrenal hyperplasia due to 21-hydroxylase deficiency in treatment monitoring.

Bina Akura, author

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Abstrak

Hiperplasia adrenal kongenital (HAK) adalah gangguan yang ditandai dengan defek pada enzim jalur steroidogenesis adrenal. Lebih dari 90% disebabkan mutasi pada CYP21A2 yang mengkode enzim 21-hidroksilase (21-OH). Monitoring tata laksana pasien HAK cukup sulit dicapai dengan menjaga keseimbangan antara overtreatment dan undertreatment.

Penelitian bersifat cross sectional dilakukan di RSCM berlangsung selama 7 bulan (Juni-Desember 2020). Pemilihan sampel dilakukan dengan consecutive sampling dengan total 142 sampel. Sampel pasien HAK sebanyak 71 pasien, serta pemilihan sampel kontrol dilakukan dengan matching jenis kelamin dan usia. Kelompok HAK dilakukan pemeriksaan kadar 17-hidroksiprogesteron (17-OHP) serta androsteron, etiokolanolon urin dan rasio androsteron/etiokolanolon urin (A/E). Uji korelasi dilakukan antara androsteron, etiokolanolon, rasio A/E dengan 17-OHP. Kelompok kontrol dilakukan pemeriksaan androsteron, etiokolanolon urin dan rasio A/E. Hasil kedua kelompok dilakukan komparasi.

Dari 71 kelompok HAK dan 71 kelompok kontrol mempunyai karakteristik dasar yang sebanding. Kadar androsteron kelompok HAK dibandingkan dengan kelompok kontrol berbeda bermakna (683,89(29,42-61061,43) vs 123,97(30,16- 16463,05) ng/mL; $p < 0,001$). Kadar etiokolanolon kelompok HAK berbeda bermakna dibandingkan dengan kelompok kontrol (235,88(12,77-78446,65) vs 70,96(12,61-17332,62)ng/mL; $p < 0,001$). Rasio A/E kelompok HAK berbeda bermakna dibandingkan dengan kelompok kontrol (2,31(0,37-40,12) vs 1,99(0,52- 5,45); $p = 0,003$). Kadar androsteron, etiokolanolon, rasio A/E mempunyai korelasi positif dengan kadar 17-OHP ($r = 0,505$; $r = 0,367$; $r = 0,313$).

Kesimpulan: Androsteron, etiokolanolon dan rasio A/E mempunyai kadar yang lebih tinggi pada pasien HAK. Androsteron mempunyai korelasi sedang, etiokolanolon dan rasio A/E mempunyai korelasi lemah terhadap 17-OHP.

.....Congenital adrenal hyperplasia (CAH) is a disorder characterized by defects in one of the enzymes of the adrenal steroidogenesis pathway. More than 90% of cases are due to mutations in CYP21A2, the gene coding for 21-hydroxylase (21-OH) enzyme. Treatment monitoring in CAH patients is quite difficult to achieve due to fine balance of overtreatment and undertreatment.

Cross sectional study was conducted in RSCM for 7 months (June-December 2020). Consecutive sampling was used with total 142 samples. There were 71 patients CAH were included in this study. Control samples were selected by matching age and sex. In CAH group, 17-hydroxyprogesterone (17-OHP), urine androsterone, etiocholanolone, and ratio androsterone/etiocholanolone (A/E) were measured. Correlations were measured between androsterone, etiocholanolone, ration A/E with 17-OHP. In control sample urine androsterone, etiocholanolone, and ratio androsterone/etiocholanolone (A/E) were also measured. These

results were compared between two groups.

In 71 CAH group and 71 control group had almost same characteristics. Androsterone level in CAH group had a significant different compared to the control group (683.89(29.42-61061.43) vs 123.97(30.16-16463.05) ng/mL; $p < 0.001$). Etiocholanolone level in CAH group had a significant different compared to the control group (235.88(12.77-78446.65) vs 70.96(12.61- 17332.62)ng/mL; $p < 0.001$). Ratio A/E n CAH group had a significant different compared to the control group (2.31(0.37-40.12) vs 1.99(0.52-5.45); $p = 0.003$). Androsterone, etiocholanolone and ratio A/E had positive correlation with 17-OHP level ($r = 0.5050$; $r = 0.367$; $r = 0.313$).

Conclusions: Androsterone, etiocholanolone, and ratio A/E had higher level in CAH subjects. Androsterone had intermediate correlation with 17-OHP, meanwhile etiocholanolone and ratio A/E had weaker correlation.