

DISERTASI

**ANALISIS GENETIK DAN FAKTOR RISIKO STATUS VITAMIN D
PADA IBU HAMIL DAN HUBUNGANNYA TERHADAP
ANTROPOMETRI BAYI BARU LAHIR**



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ABSTRAK

ANALISIS GENETIK DAN FAKTOR RISIKO STATUS VITAMIN D PADA IBU HAMIL DAN HUBUNGANNYA TERHADAP ANTROPOMETRI BAYI BARU LAHIR

Arif Sabta Aji

Defisiensi vitamin D merupakan salah satu masalah utama kesehatan masyarakat di dunia dan ditemukan hampir pada semua daur kehidupan. Penelitian tentang peran vitamin D dalam pertumbuhan janin telah banyak dibahas dengan hasil yang bertentangan. Namun, penelitian tersebut masih terbatas di Indonesia. Penelitian ini bertujuan untuk menganalisis hubungan status vitamin D ibu selama hamil dengan antropometri bayi baru lahir berdasarkan faktor risiko status vitamin D, kadar IGF-I, dan polimorfisme gen yang berhubungan dengan sintesis dan metabolisme vitamin D.

Penelitian ini merupakan penelitian observasional dengan desain *cross-sectional* pada 180 subjek penelitian ibu hamil sehat. Data dikumpulkan mulai dari trimester pertama (T1) sampai proses persalinan. Data antropometri bayi baru lahir diperiksa mulai dari berat badan, panjang badan, dan lingkar kepala. Kadar serum 25-hidroksivitamin D (25OHD) dan kadar IGF-I diperiksa saat trimester ketiga (T3) menggunakan *Enzyme-linked Immunosorbent Assay* (ELISA). Analisis genetik dilakukan dengan metode *PCR-KASP*.

Hasil penelitian memperlihatkan bahwa ada peningkatan kadar 25(OH)D selama kehamilan. Faktor risiko yang terkait dengan status vitamin D ibu hamil T1 adalah status tidak kerja, lama aktivitas di luar ruangan kurang dari satu jam, dan tidak ada konsumsi suplemen sebelum kehamilan. Ada hubungan yang signifikan dalam gen *VDR* (rs7975232), *CYP2R1* (rs12794714), dan *GC* (rs22282679) dengan rerata kadar 25(OH)D selama kehamilan. *Weighted OR-GRS* secara signifikan berhubungan dengan rerata kadar 25(OH)D selama kehamilan. Status vitamin D selama kehamilan memiliki hubungan yang signifikan dengan kadar IGF-I di T3. Hasil penelitian ini tidak menemukan hubungan antara status vitamin D dan antropometri bayi baru lahir.

Kesimpulan dari penelitian ini adalah terdapat prevalensi status defisiensi vitamin D pada ibu hamil T1 yang tinggi di Sumatra Barat, terdapat hubungan antara SNPs dari gen yang berperan pada sintesis dan metabolisme vitamin D dengan kadar 25(OH)D selama kehamilan dan terdapat hubungan antara GRS dan kadar 25(OH)D selama kehamilan. Status vitamin D selama kehamilan tidak berhubungan dengan antropometri bayi baru lahir. Namun, status vitamin D memiliki hubungan dengan kadar IGF-I pada T3 kehamilan.

Kata Kunci: Status vitamin D, kehamilan, SNPs, IGF-I, skor risiko genetik, antropometri bayi baru lahir

ABSTRACT

ANALYSIS OF GENETIC AND RISK FACTORS OF VITAMIN D STATUS IN PREGNANT WOMEN AND ITS ASSOCIATION WITH NEWBORN ANTHROPOMETRY MEASUREMENTS

Arif Sabta Aji

Vitamin D deficiency is one of the main public health problems in the world and affects in almost all life cycles. Research on the role of vitamin D in foetal growth has been widely discussed with conflicting results. However, the research is limited in Indonesian population. This study aimed to analyze the relationship between maternal vitamin D status during pregnancy and newborn anthropometry outcomes based on risk factors for vitamin D status, IGF-I level, and genetic polymorphisms which associated with vitamin D synthesis and metabolism pathway.

This study was an observational cross-sectional study on 180 subjects in healthy pregnancy. Data was collected from the first trimester (T1) to the delivery process. Newborn anthropometric measurement such as birth weight, birth length, and head circumference were assessed. Serum 25-hydroxyvitamin D (25OHD) and Insulin-like Growth Factor I (IGF-I) concentration were measured at the third trimester (T3) using Enzyme-linked Immunosorbent Assay (ELISA). Genotype analysis was carried out using PCR-KASP.

The result of this study showed that there was a significant increase of maternal serum 25(OH)D concentration during pregnancy. There were risk factors associated with vitamin D deficiency in T1 such as non-work status, duration of outdoor activities which less than one hour, and not taking supplements before pregnancy. There was a significant association of *VDR* (rs7975232), *CYP2R1* (rs12794714), and *GC* (rs22282679) with a mean concentration of 25(OH)D during pregnancy. Weighted OR-GRs were significantly associated with the mean concentration of 25(OH)D during pregnancy. Vitamin D status during pregnancy had a significant association with serum IGF-I concentration in T3. There was no significant association between vitamin D status during pregnancy and newborn anthropometry outcomes.

The conclusions of this study were that the prevalence of vitamin D deficiency in T1 was high in West Sumatra, there was an association between SNPs from genes that regulate synthesis and metabolism of vitamin D and serum 25(OH)D concentration during pregnancy and there was an association between GRS and serum 25(OH)D concentration during pregnancy. There was no association between maternal vitamin D status and newborn anthropometry. However, maternal vitamin D status was associated with IGF-I concentration in T3 of pregnancy.

Keywords: Vitamin D status, pregnancy, SNPs, IGF-I, genetic risk scores, newborn anthropometry