

Variasi sekuen DNA promoter Gen AMH (anti mullerian hormone) analisisnya terhadap kadar serum AMH dan jumlah folikel antral pada pasien SOPK (Sindrom Ovarium Polikistik) = DNA sequence variation AMH anti mullerian hormone gene promoter analysis of serum AMH level and number follicle antral in PCOS (Policystic Ovary Syndrome)

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Abstrak

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LATAR BELAKANG: Anti Mullerian Hormon (AMH) adalah anggota dari golongan Transforming Growth Factor- yang berperan dalam pengaturan folikulogenesis pada reproduksi wanita. Peningkatan kadar AMH 2 sampai 3 kali dijumpai pada pasien SOPK (Sindrom Ovarium Polikistik) daripada wanita dengan ovulasi normal. Pada penelitian ini dideteksi varian sekuen disepanjang daerah promoter gen AMH. Adanya variasi promoter gen AMH diduga mempengaruhi proses transkripsi gen AMH yang selanjutnya berimplikasi pada pembentukan protein AMH. Apabila terjadi gangguan pada pembentukan protein AMH maka akan berpengaruh terhadap kadar protein tersebut di dalam darah.

BAHAN DAN CARA KERJA: Sampel penelitian ini berjumlah 114 pasien yang terdiri dari 60 pasien SOPK dan 54 pasien non SOPK (Kontrol). Kadar AMH dan Jumlah folikel antral didapatkan dari data rekam medik pasien Klinik IVF Yasmin, RSCM Kencana Jakarta. Analisis molekuler dan genotyping dilakukan dengan teknik PCR dan sekuensing kemudian dilanjutkan dengan analisis bioinformatika.

HASIL : Dari penelitian ini ditemukan 60 titik varian mutasi promoter gen AMH. Jenis varian mutasi terbesar yang ditemukan adalah -674 G/A (100 %), -245 C/CT (88,2 %), dan -444 A/G (17,9 %) dari seluruh sampel. Berdasarkan hasil uji Wilcoxon Signed Ranks, pada kelompok SOPK ditemukan jumlah mutasi yang terjadi berpengaruh secara bermakna terhadap kadar AMH dan jumlah folikel antral ( $p < 0,05$ ). Pada kelompok kontrol ditemukan bahwa jumlah mutasi tidak berpengaruh secara bermakna terhadap kadar AMH ( $p > 0,05$ ), tetapi berpengaruh secara bermakna terhadap jumlah folikel antral ( $p < 0,05$ ). Ditemukan 60 titik varian pada promoter gen AMH. Jumlah mutasi pada promoter gen AMH berpengaruh terhadap kadar AMH dan jumlah folikel antral pada SOPK. Mutasi pada titik -674 G/A merupakan titik mutasi baru yang belum pernah dilaporkan oleh NCBI, ditemukan pada seluruh subyek penelitian baik kelompok SOPK maupun non SOPK.

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<b>ABSTRACT</b><br>

INTRODUCTION : Anti-Mullerian Hormone (AMH) is a member of the Transforming Growth Factor-group which plays an important role in the regulation of the female reproductive folliculogenesis. A 2-3 fold increase in AMH levels was found in patients with PCOS (Polycystic Ovary Syndrome) compared to women with normal ovulation. This study detected sequence variants in the AMH gene promoter region. The AMH gene promoter variation is thought to affect AMH gene transcription process implicated in the formation of proteins. In the event of disruption in the formation of these AMH proteins, the levels of these proteins in the blood will be affected. The purpose of this study was to detect variants of AMH gene promoter sequences.

**MATERIALS AND METHODS:** The sample size was 114 patients consisting of 60 PCOS patients and 54 non-PCOS patients as control. The AMH levels and antral follicle number obtained from the patients' medical records of the Yasmin IVF Clinic, RSCM Kencana Hospital, Jakarta. Molecular analysis and genotyping were performed by PCR and sequencing was followed by bioinformatics analysis.

**RESULTS:** There were 60 point mutations in the AMH gene promoter variants. The highest variant types of mutations found was -674 G/A (100%), followed by -245 C/CT (88.2%), and -444 A/G (17.9%) in the entire sample. Based on the results of the Wilcoxon Signed Rank test, the number of mutations in the PCOS group were significant to effect the serum AMH level and the antral follicle number ( $p < 0.05$ ). In the control group, the number of mutations had no significant effect on the levels of AMH ( $p > 0.05$ ), but significantly affected the number of antral follicles ( $P < 0.05$ ). There were 60 point variances in the AMH gene promoter. The number of mutations in the gene promoter affected serum AMH levels and the number of antral follicles in PCOS. A new point mutation was found in all subjects at position -674 G/A, which have not been reported by the NCBI.; **INTRODUCTION :** Anti-Mullerian Hormone (AMH) is a member of the Transforming Growth Factor- group which plays an important role in the regulation of the female reproductive folliculogenesis. A 2-3 fold increase in AMH levels was found in patients with PCOS (Polycystic Ovary Syndrome) compared to women with normal ovulation. This study detected sequence variants in the AMH gene promoter region. The AMH gene promoter variation is thought to affect AMH gene transcription process implicated in the formation of proteins. In the event of disruption in the formation of these AMH proteins, the levels of these proteins in the blood will be affected. The purpose of this study was to detect variants of AMH gene promoter sequences.

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