

## Single nucleotide polymorphisms of follicle-stimulating hormone receptor promoter and their impacts to the promoter activities

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### Abstrak

Respon wanita usia reproduksi bervariasi terhadap stimulasi FSH eksogen. Salah satu penyebab variasi tersebut adalah perbedaan genotip akibat adanya polimorfisme pada ekson 10 gen reseptor FSH. Untuk mengetahui lebih lanjut apakah daerah promotor inti gen reseptor FSH juga polimorfik dan apakah polimorfisme tersebut mempengaruhi aktivitas promotor, dilakukan skrining polimorfisme promotor gen reseptor FSH pada 262 wanita yang mengikuti program IVF/ICSI, diikuti uji fungsional untuk mengetahui pengaruh polimorfisme terhadap aktivitas promotor. Hasil penelitian menunjukkan bahwa daerah promotor inti gen reseptor FSH polimorfik. Ditemukan lima SNPs pada posisi -29, -37, -114, -123 dan -138 di samping ditemukannya variasi jumlah basa adenin. Polimorfisme pada posisi -123 menurunkan aktivitas promotor secara bermakna, sebaliknya polimorfisme pada posisi -37 dan -138 meningkatkan aktivitas promotor secara bermakna, sedangkan polimorfisme pada posisi -29, -114 dan pemendekan basa adenin tidak mempengaruhi aktivitas promotor secara bermakna. Perbedaan aktivitas promotor akibat polimorfisme ini pada akhirnya sangat memungkinkan merubah sensitivitas ovarium terhadap FSH. (Med J Indones 2004; 13: 205-14)

*Women of reproductive ages are varies in their responses to exogenous FSH stimulations. The difference of FSHR genotype due to the polymorphisms in exon 10 is one of its significant factors. To know further whether the core promoter of FSHR is also polymorphic and to know whether those polymorphisms influence the promoter activity, we did polymorphism screening of FSHR promoter to 262 women undergoing IVF/ICSI, followed by functional study to know the impact of polymorphisms to the promoter activity. This study indicated that the core promoter of human FSHR is polymorphic. We found five SNPs at positions -29, -37, -114, -123 and -138 in addition to the variety number of adenines. Polymorphism at position -123 significantly decreased the promoter activity, in contrast, polymorphism at position -37 and -138 significantly increased the promoter activity, whereas polymorphism at position -29, -114 and short adenines stretch did not significantly influence the promoter activity. The differences of the promoter activities due to polymorphisms might change the ovarian sensitivity to FSH. (Med J Indones 2004; 13: 205-14)*