

## FSHR gene Thr307Ala and Asn680Ser polymorphisms in infertile men: an association study in North China and meta-analysis

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**ABSTRACT.** Male infertility is a complex multifactorial and polygenic disease, and genetic factors play an important role in its formation and development. Recently, the association between follicle stimulating hormone receptor (FSHR) gene polymorphisms and male infertility risk has attracted widespread attention due to the unique biological functions of FSH. The aim of this study was to further explore the associations between the Thr307Ala and Asn680Ser polymorphisms of the FSHR gene and male infertility. A case-control study of 212 infertile and 164 fertile men from North China was performed. FSHR polymorphism genotypes were obtained through direct DNA sequencing. A metaanalysis was also performed. In the single-site association analysis, no significant associations were identified between FSHR Thr307Ala and As n680Ser polymorphisms and male infertility (P > 0.05). However, we found that the combined genotypic frequency of Thr/Ala + Asn/ As was higher in infertile patients than in controls (6.6 vs 1.8%; odds ratio (OR) = 3.795; 95% confidence interval (CI): 1.072-13.434, P = 0.027). In the meta-analysis, there was also no evidence of FSHR polymorphism (rs 6165 and rs 6168) association with male infertility (P>0.05). However, we found that the combined genotypes Thr/Thr + Asn/Asn had an increased risk of male infertility (OR = 1.238; 95%CI: 1.001-1.537, P = 0.049). Our studies further confirmed reports that there were no significant associations between the *FSHR* Thr307Ala and Asn680Ser polymorphisms and male infertility risk. However, a combined FSHR genotype showed significant association with male infertility.

**Key words:** Male infertility; Idiopathic infertility; Meta-analysis; Follicle-stimulating hormone receptor; Single nucleotide polymorphisms