Background: Recurrent pregnancy loss (RPL), affecting 1-2% of couples, is a multifactorial disorder as both genetic and environmental factors are involved. The aim of this study was to determine association of single nucleotide polymorphisms (SNPs) located on estrogen receptor alpha gene (ESR1); -397C/T and -351A/G in intron 1, and estrogen receptor beta gene (ESR2); +1082G/A in exon 5, +1730G/A in exon 8, and rs1256030 C/T in intron 2 with the risk of RPL in Iranian women.

Material and methods: In this study, 250 women with a history of three or more consecutive pregnancy losses before 20th week of gestation and 105 healthy women with at least two live births and no history of pregnancy loss were included. PCR-RFLP was employed and the data was statistically analyzed using Mann-Whitney U test to explore the relationship between the investigated SNPs and RPL.

Results: A statistically significantly higher frequency of GA genotype (+1082G/A) was detected in women with RPL compared to controls (P < 0.05). There were no significant differences in other investigated polymorphisms on ESR1 and ESR2 genes in subjects. In addition no association was found between the analyzed genotypes and hormonal profile of Follicle Stimulating Hormone, Luteinizing Hormone and Estradiol in investigated subjects.

Conclusion: Our findings may suggest the involvement of some SNPs which are located on functional region of ESR2 gene as a risk factor for RPL in Iranian women.

Key word: estrogen receptor, polymorphism, recurrent pregnancy loss.