MTHFR C677T Polymorphism and Recurrent Early Pregnancy Loss Risk in North Indian Population

1. **Rohini R. Nair, MSc**[1]
2. **Anuradha Khanna, MD**[2]
3. **Kiran Singh, PhD**[1]⇓

1. [1]Department of Molecular & Human Genetics, Banaras Hindu University, Varanasi, India
2. [2]Department of Obstetrics & Gynaecology, Institute of Medical Sciences, Banaras Hindu University, Varanasi, India

1. Kiran Singh, Department of Molecular & Human Genetics, Banaras Hindu University, Varanasi 221005, India Email: singhk4@rediffmail.com

Abstract

Recurrent early pregnancy loss (REPL) is a multifactorial disorder as both genetic and environmental factors contribute to the development of disease. Folate metabolism is an important mechanism to ensure proper fetal growth. Hyperhomocysteinemia leads to a number of disorders and REPL is one of them. In a case–control study DNA from 106 cases with the history of 3 or more REPL and 140 healthy fertile controls with successful pregnancy outcomes were genotyped for C677T single-nucleotide polymorphism (SNP) of the *MTHFR* (methylenetetrahydrofolate reductase) gene through polymerase chain reaction–restriction fragment length polymorphism (PCR-RFLP), which was further confirmed by sequencing. Allele frequencies of REPL cases were compared with healthy controls and a statistically significant association was found between REPL and the mutant T allele (χ^2 = 8.786, odds ratio [OR] = 2.20, 95% confidence interval [CI] = 1.323-3.9658, *P* = .003). The genotype frequencies of SNP C677T also differ significantly between these 2 groups (χ^2 = 8.237, *P* = .016). The OR for heterozygous CT in the REPL versus controls is 1.9591 (95% CI = 1.0285-3.7318,*P* = .04). The OR for TT homozygous is 6.3009 (95% CI = 1.2065, *P* = .02). Combined odds ratio of CT and TT against the control has been calculated as 2.2194 (95% CI = 1.2029-4.0952, *P* = .02) which is also significant. Thus the present study clearly indicates that homozygosity and heterozygosity for the *MTHFR* C677T polymorphism confer a 6.3009- and 1.9591-fold increased risk of idiopathic REPL, respectively.