The etiology underlying neural tube defects (NTDs) is not fully understood and is believed to involve a complex milieu of genetic and environmental factors. The A1298C polymorphism in the methylenetetrahydropholate reductase gene (MTHFR) has been associated with mild risk for NTDs. In this study, the genotype distribution of the MTHFR gene A1298C polymorphism and the levels of serum homocysteine, vitamin B12, and folate were evaluated in 33 children with NTDs, their mothers, and 46 healthy controls. Genotyping of the A1298C polymorphism was performed by real-time polymerase chain reaction. The A and C allele frequencies in children with NTDs and their mothers were similar to controls (P = 0.160). The 1298AA and 1298CC genotype frequencies (P = 0.551 and 0.062, respectively) in children with NTDs and their mothers did not differ from controls. On the other hand, the 1298AC genotype frequencies in children with NTDs and their mothers were significantly different from controls (P = 0.025). The genotype frequency of 1298AC was lower in children with NTDs than in controls. There was no significant association between clinical distribution of NTDs and 1298AA/AC/CC genotypes (P > 0.05). Serum vitamin B12 levels were higher in children with NTDs than their mothers and controls (P = 0.001). There were no differences among serum homocysteine and folate levels in all groups (P = 0.494 and 0.141, respectively). Both genetic and nutritional factors are important in the etiology of NTDs. Thus, the A1298C polymorphism cannot be regarded as a major risk factor for NTDs [1)].

1)
Yildiz SH, Ozdemir Erdogan M, Solak M, Eser O, Arıkan Terzi ES, Eser B, Kocabaş V, Aslan A. Lack of association between the methylenetetrahydropholate reductase gene A1298C polymorphism and neural tube defects in a Turkish study group. Genet Mol Res. 2016 Jun 3;15(2). doi: 10.4238/gmr.15028051. PubMed PMID: 27323133.