Az MTHFR-génpolimorfizmusok szerepe a folsav élettani hatásainak alakulásában a várandósság alatt

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Összefoglalás
A metilén-tetrahidrofolát reduktáz (MTHFR) enzim a biológiailag aktív folsav,- az 5-metil tetrahidrofolát kialakulását katalizálja a szervezetben. Az MTHF C677T mutációja csökkent enzimaktivitást eredményez, melynek következtében a homocisztein remetilációjához szükséges 5-MTHF biológiai elérhetősége lecsökken. A mutáció ezért hiperhomociszteinaemiát okozhat, ami potenciálisan káros hatással lehet a fejlődő embrióra várandósság alatt. A magas homocisztein koncentráció feltehetőleg a metilációs folyamatokat változtatja meg az embrionális fejlődés során. Az MTHFR mutáció így más környezeti és genetikai faktorokkal kombinálódva megnövelheti a velőcső záródási rendellenességek és a spontán vetélés kialakulásának valószínűségét is. Jelentősebb kockázatot jelent, ha az anya és a magzat is TT genotípússal rendelkezik illetve a folsav ellátása elégtelen. A mutációt hordozóknál megnövelt dózisú folsav supplementáció ajánlott az egészséges magzati fejlődés érdekében.
Kulcsszavak: velőcső-záródási rendellenesség, folsav, MTHFR-génpolimorfizmus, homocisztein

The association between MTHFR polymorphisms and folate metabolisms during pregnacy

Abstract
The methylenetetrahydrofolate reductase (MTHFR) catalyzes the conversion of 5,10 methylenetetrahydrofolate to 5-methyltetrahydrofolate (5-MTHF) required for the conversion of homocysteine to methionine. The C677T polymorphisms results in lower specific activity of MTHFR associated with reduced bioavailability of 5-MTHF. The individuals with TT genotype are therefore predisposed to mild hyperhomocysteinaemia. Elevated level of homocysteine might cause decreased fetal viability due to the altered metylation reactions in the developing embryo. Thus the MTHFR mutation- combined with other genetic and environmental factors- can increase the risk of neuronal tube defects (NTD) and spontaneous abortion. In addition, the risk for NTD can be higher if both mother and child are homozygous for the mutation. Women with MTHFR mutation might have higher needs for folate supplementation to reduce the risk of folic-acid related pregnancy complications.
Keywords: neuronal tube defects, folate, MTHFR gene polimorphism, homocysteine

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