Relationship between C677T Methylenetetrahydrofolate Reductase Gene Polymorphism and Homocysteine in Cerebral Palsy

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Abstract

Objective: To observe the relationship between methylenetetrahydrofolate reductase (MTHFR) C677T gene polymorphism and homocysteine levels in cerebral palsy (CP) children.

Methods: This cross-sectional study was conducted in several hospitals, school for children with special needs, and rehabilitation centers in Bandung from March to November 2014, on children with CP aged 4–14 years who met the inclusion criteria. Genotyping was performed using polymerase chain reaction (PCR)-restriction fragment length polymorphism (RFLP) and direct sequencing. Homocysteine serum level was measured using chemiluminescent microparticle immunoassay (CMIA) method. Statistical analysis was conducted using t test.

Results: In this study, 150 CP children had MTHFR C677T gene polymorphism with a frequency of 18%, consisting of TT homozygotes (4%), CT heterozygotes (14%), and T allele (11%. The mean serum level of homocysteine in CP with C677T MTHFR gene polymorphism was 8.22 (±1.89) μmol/L, higher than those without polymorphism (p=0.046).

Conclusions: A relationship between MTHFR C677T gene polymorphism and homocysteine level in children with cerebral palsy is found in this study.

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Introduction

Cerebral palsy (CP) is a neurodevelopmental disorder that is most frequently found and one of the main causes of physical disability in children. The worldwide prevalence of CP is 2–3/1000 live births.[1] In Indonesia there are currently no data on the prevalence of CP.

CP is a disease with multifactorial causes; 70–80% of all cases occur during the prenatal period and the rest of the cases occur during perinatal or postnatal period. It is currently considered that genetic disorders play an important role in the occurrence of CP. These genetic disorders are also found in some other developmental disorders, such as intelligence disorders and autism spectrum disorders.[2,3] A number of studies were conducted to support this statement, such as studies on CP-related genes as well as studies on the presence of single nucleotide polymorphisms (SNPs).[4,5] In this respect, Callaghan *et al.*[6] had conducted a systematic literature review and discovered that there were approximately 22 studies related to the relationship of SNPs and CP. One of the candidate genes in this relation with CP is methylenetetrahydrofolate reductase (MTHFR). However, results of studies on the relationship between CP and genetic factors were still unsatisfactory due to the limitation in subjects, ethnic differences, and inadequate control. A study conducted in China reported that there is a significant relationship between MTHFR C677T gene polymorphism and CP

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