INTRODUCTION

The methylenetetrahydrofolate reductase gene mutation, known as MTHFR is a well-known DNA repair enzyme. Approximately 10-15% of the US population have a clone of the MTHFR gene mutation (Rady et al., 1990). MTHFR is critical to the proper functioning of DNA. Mutations in the MTHFR gene can lead to neurological disorders, such as hypotonia and optic neuropathy.

METHODS

The two most common forms of the mutation are C677T and A1298C. These mutations can be detected through DNA analysis. The C677T mutation is present in 10% of the general population and in 33% of individuals with a history of miscarriage. The A1298C mutation is present in 10% of the general population and in 20% of individuals with a history of miscarriage (Rozen R. Methylenetetrahydrofolate reductase: a link between folate and riboflavin? Am J Clin Nutr 1991; 54: 185-91).

RESULTS

The most frequent articles were the following: (1) Methyl-tetrahydrofolate deficiency is a disorder of homocysteine metabolism. (2) MTHFR and HYPOTONIA, which showed 50 articles and HOMOCYSTEINE and VISION, which showed 4 publications. In regard to hypotonia, the query HYPOTONIA AND VISION found 17 articles, and HOMOCYSTEINE AND VISION showed 3 publications.

CONCLUSION

More serious under-diet are related to the MTHFR gene defect. In addition to homocysteine, in the diagnosis and treatment of the disease that has been here. Based on a study of the human research, it is more important that the patient and/or family determine to be harbored with other factors.

After reading the above description of the relationship of hypotonia to vision disorders, Behcet's disease, primary open angle glaucoma, primary closed angle glaucoma, age related macular degeneration, and Leber Hereditary Optic Neuropathy, vision related symptoms may be present in the child with MTHFR.

REFERENCES


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