Cureus

Open Access
Abstract

Cureus

Further Analysis of Multicentre Cystathionine Beta Synthase Deficiency Thrombosis Data and Metabolic Pathways Suggests Potentially Better Treatment via Improved Cysteine Supplementation, Diet, Antioxidant Supplementation, Follow-up and Testing...

David J. Vance [1]

1. Independent Consultant, Windsor, AUS

Corresponding author: David J. Vance, davidjmvance@hotmail.com

Categories: Public Health
Keywords: cystathionine beta synthase, homocystinuria, methionine, cysteine, homocysteine, diet, vitamin c, fmd

How to cite this abstract
Vance D J (January 01, 2017) Further Analysis of Multicentre Cystathionine Beta Synthase Deficiency Thrombosis Data and Metabolic Pathways Suggests Potentially Better Treatment via Improved Cysteine Supplementation, Diet, Antioxidant Supplementation, Follow-up and Testing.... Cureus 9(1): a302

Abstract

Background: Homozygous or compound heterozygous Cystathionine-beta-synthase deficiency (CBS--) may result in thrombosis. Treatment has included various combinations of: low-methionine diets, cystine (cystine dimer)-enriched amino acid supplementation, vitamin B6, folic acid, vitamin B12 and betaine. Treatment compliance and outcomes even in the most-developed countries are mostly sub-optimal and variable, and the differing theoretical metabolic ramifications due to differing treatments have not been well addressed. The aim of this work was to further analyse the thrombosis events data of Yap et al (2001/2003), and to compare these with the rate of thrombosis in the general population, and to examine the theoretical significance of the metabolic pathways affected by CBS-- and its treatments, and so find any potential improvements in treatments, considering also less-developed areas.

Methods: Yap et al's (2001/2003) data of the thrombosis outcomes of five major (CBS--)-treating centers: in Dublin, Sydney, Nijmegen, Manchester and London; were statistically compared with outcomes predicted by Mudd et al's (1985) untreated natural history outcomes, and then Dublin versus the others; these rates were then compared with those of general populations; and treatments were examined regarding their theoretical metabolic ramifications.

Results: There were less thrombosis outcomes (P<.05) in the treated and followed CBS-- patient groups of each of the five centers, even when considered singly, than that expected in the absence of treatment by reference to the natural history data of Mudd et al (1985), but the reduction was less than half that claimed by Yap et al, and the remaining level of thrombosis is roughly 10 times that of the general population. The thromboses outcome (nil) of the Dublin group is better than that of the other four groups, but only at P ~ 0.16 with the other four groups combined, or P = 0.14 to 0.23 singly. Treatment regimens differ, including in the sub-optimality of metabolic outcomes, due not only to CBS-- but also its treatments.

Conclusions: It seems likely that substantial improvements in the treatment of CBS-- may be achieved through

Open Access
Abstract
Published 01/01/2017

1. Cysteine supplementation (preferably on its own rather than in whole-diet formula mixtures), in accord with various uses of homocysteine-lowering nutrients other than VitB6, which have various effects on the metabolism of homocysteine to cysteine.
2. Better use of low-methionine, high-fruit and vegetable whole-food diets.
3. Supplementation with vitamin C and other antioxidants.
4. Better cultivation of patient compliance.
5. Testing for Factor 5 Leiden and prothrombin C20210A mutations.

Full article: http://gjmedph.com/uploads/O6-Vo6No2.pdf