Disorders of folate metabolism

Vitamin B9 (folic acid)

- Folate (pteroylglutamic acid) is found in leafy vegetables, legumes, liver
- Pteroylglutamic acid consists of pteridine, p-aminobenzoic acid and glutamic acid
- Reduction to **dihydrofolate** (DHF) and **tetrahydrofolate** (THF) occurs in folate metabolism
- It serves as a transporter of single-carbon residues, which is essential for the endogenous formation of methionine, thymidylate (dTMP) and two intermediates in purine synthesis.

Hereditary folate malabsorption

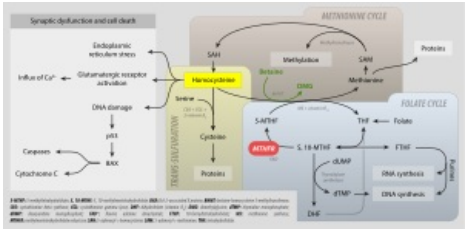
- Severe megaloblastic anemia, progressive neurological disorders
- Folate deficiency syndrome in the brain with reduced levels of folate transporter in the brain
- Treatment with high doses of folate orally or lower parenterally

Glutamate formimino transferase deficiency

- Histidine catabolism is associated with the transfer of the formimino group to THF with the concomitant release of ammonia
- It has two catalytic activities as **glutamate formiminotransferase** and as **formiminotetrahydrofolate cyclodeaminase**
- Psychomotor retardation, megaloblastic anemia
- Hyperhistinemia and histidinuria develop

Methylenetetrahydrofolate reductase deficiency (MTHFR)

- Methyl-THF is a methyl donor for the conversion of homocysteine to methionine and the MTHFR deficiency results in an increase in total plasma homocysteine concentration and a decrease in methionine concentration leading to developmental delay, microcephaly, convulsions, hyperhomocysteinemia



MTHFR metabolism

Links

- Vitamin B9
- Disorders of cobalamin metabolism
- Disorders of sulfur amino acid metabolism

Source

- FERNANDES, John. *Diagnostika a léčba dědičných metabolických poruch.* 1. vydání. Praha : Triton, 2008. s. 576-580. ISBN 978-80-7387-096-6.
- MURRAY, Robert K., Daryl K. GRANNER a Peter A. MAYES, et al. *Harperova biochemie.* 4. vydání. Jinočany : H & H, 2002. 872 s. ISBN 80-7319-013-3.