# Guide to Severe MTHFR

(Severe Methylene tetrahydrofolate reductase)

## Teachers and School Nurses

## What is severe MTHFR

Severe MTHFR is a very rare genetic condition and it occurs when the body is unable to process certain amino acids (building blocks of protein) properly. Severe MTHFR causes a buildup of the Homocysteine (HCY) and a deficiency in the amino acid, methionine. In severe MTHFR, the enzyme methylene tetrahydrofolate reductase does not work at all. This is one of the disorders that is called a methylcobalamin disorders since the conversion of HCY to methionine requires vitamin B12 (cobalamin) and conversion of the vitamin folate to methylfolate (which is the actual role of the enzyme methylene tetrahydrofolate reductase in this process).

## Why have I heard of MTHFR when the severe form is so rare?

There are two common variations of MTHFR which, in most populations, at least 30% of people have. It has made the news since at one time it was associated with many diseases. In most cases, the common variants of MTHFR do not cause problems (if they did, we would see these problems much more commonly given that so many people have these variants or in this case polymorphisms). Rarely do people with the common variants have problems with higher HCY, but usually this is due to vitamin B12 deficiency or folate deficiency and so supplementation can help.

## What happens?

Normally, a metabolic process occurs in our body where homocysteine (HCY) is converted into another amino acid, methionine. A genetic mutation prevents this process from occurring and a dangerous build-up of HCY. Supplementation and medication are needed to assist this process and maintain close to normal levels of HCY and methionine. Vitamin B12 (sometimes by injection) and other medications are needed to lower HCY, increase methionine and avoid harmful effects. High HCY and low methionine levels are harmful to the eyes, bones, blood vessels and the nervous system.

## High HCY levels may cause:

- Feeding Difficulties
- Vision Problems
- Seizures
- Poor Muscle tone and coordination
- Developmental Delay
- Intellectual disabilities
- Neurological abnormalities
- Blood clots or strokes

## How can I help?

**Teach** your student as you would anyone else. HCY and MMA levels can fluctuate. Your student with HCU+MMA may need additional time or attention to keep pace with the classroom. Some may have visual impairment and need visual support.

**Help** to ensure your student takes their medication(s). Peer pressure may cause children to be difficult about taking their medication, or avoid it.

**Communicate** with parents and ask questions. Since you spend a fair amount of time with your student, you may be the first to notice issues related to HCU. Successful HCU management will rely on both parents and school staff communicating with each other.

## Helpful Tips

- Treat them the same as your other students. Your student is not sick and shouldn't be treated as such. If they follow their medication regimen they can be just as successful as their classmates.
- Do not let severe MTHFR define your student. Establishing a sense of self outside of their disorder is a crucial part of self acceptance.
- Patients can't feel when their HCY levels are high (unlike a diabetic who can feel when their insulin is low/high). Regular blood draws are required to manage medication and mitigate negative effects. Symptoms usually occur after a prolonged period of high homocysteine levels. Levels increase gradually, they do not spike suddenly.
- Individuals with severe MTHFR can have strokes. If you are concerned that your student is actively having a stroke (symptoms include but this is not an extensive list: sudden confusion, loss of skills, difficulty communicating, one-sided weakness or facial droop), please seek immediate medical attention. If you feel that your student has had a stoke (in the recent or not so recent past), promptly discuss this with their parent.

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## Educational Accommodations

Children with severe MTHFR may experience challenges at school. These plans are available to make sure they have the best chance of succeeding in the classroom.

### Section 504 Plan:

A Section 504 Plan assists in establishing accommodations that help safeguard and ensure that a child with severe MTHFR has the same learning opportunities as other students in the classroom.

### Individual Education Plan (IEP):

An IEP is a written statement of an educational program designed to meet a child's individual needs. An IEP sets reasonable and attainable learning goals for a child with severe MTHFR.

Accommodations might include:

⇒ Preferential seating

⇒ Modified textbooks or audio-visual materials

⇒ Oral test and visual aids

⇒ Easy access to bathroom and/or nurse

⇒ Accommodations for fine and gross motor activities

⇒ Assisting with self-help skills

⇒ Access to assistive technology

## Additional Services

Because the whole body impact severe MTHFR may have, some patients may need additional services.

⇒ Speech Therapy

⇒ Occupational Therapy

⇒ Physical Therapy

⇒ Reading help

⇒ Counseling, friendship groups and help with social skills (if needed)

⇒ Protection from bullying



To learn more about severe MTHFR please visit: https://hcunetworkamerica.org

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Additional Resources:

- NIH | Genetics Home Reference—https://ghr.nlm.nih.gov/gene/MTHFR#
- E-HOD—http://www.e-hod.org/file/4146/remethylation_EN_final.pdf