

## What is MMA+HCU?

MMA+HCU is a group of rare genetic conditions that cause both Methylmalonic Acidemia (MMA) with Homocystinuria (HCU). They are usually referred to as Cobalamin disorders or cobalamin processing disorders. All these conditions lead to the body not being able to use vitamin B12 (cobalamin) properly and so the body cannot process certain fats and proteins appropriately. This results in the build-up of harmful levels of homocysteine (HCY) and methylmalonic acid (MMA) and a decrease in the amino acid, methionine. The more common disorders are Cobalamin (Cbl) C and CblD deficiency. More rare disorders include CblF, CblJ, CblX, and transcobalamin II deficiency.

## What happens?

Normally, our body uses vitamin B12 (Cobalamin) for two major processes. One occurs in our body where homocysteine (HCY) is converted into another amino acid, methionine. The other is where some amino acids make methylmalonyl-CoA which then be into succinyl-CoA. If these processes do not occur, HCY and MMA increase and can harm the eyes, skeleton, blood vessels and central nervous system. A genetic mutation prevents cobalamin from being appropriately processed, leading to these disorders and a dangerous buildup of HCY and MMA. Medications are needed to assist this process and maintain close to normal levels of HCY and MMA to decrease/avoid harmful effects.

## 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

## Possible Medication Side Effects

- Bruising
- Circular redness for injection site
- Stomach upset
- Unpleasant body odor/breath (often described as "fishy")
- Nausea



## 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 the diet they can be just as successful as their classmates.
- ✎ Do not let your child's disorder define them. Establishing a sense of self outside of their disorder is a crucial part of self-acceptance.
- ✎ Patients can't feel when their HCY and MMA 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 HCY or MMA levels. Levels increase gradually, they do not spike suddenly.
- ✎ Individuals with MMA+HCU 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 stroke (in the recent or not so recent past), promptly discuss this with their parent.

## Educational Accommodations

Children with MMA+HCU 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 MMA+HCU 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 MMA+HCU.

Accommodations might include:

- ⇒ Preferential seating
- ⇒ Modified textbooks or audio-visual materials
- ⇒ Low Vision Accommodations/ 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 MMA+HCU may have, some patients may need additional services.

- ⇒ Low Vision Therapy
- ⇒ Speech Therapy
- ⇒ Occupational Therapy
- ⇒ Physical Therapy
- ⇒ Reading help
- ⇒ Counseling, friendship groups and help with social skills (if needed)
- ⇒ Protection from bullying



This information is not intended to take the place of medical advice or care you receive from your health care professional and intended for information purposes only.

To learn more about MMA+HCU please visit: <https://hcunetworkamerica.org>

Additional Resources:

- NIH | Genetics Home Reference—<https://ghr.nlm.nih.gov/condition/methylmalonic-acidemia-with-homocystinuria#>
- E-HOD—[http://www.e-hod.org/file/4146/remethylation\\_EN\\_final.pdf](http://www.e-hod.org/file/4146/remethylation_EN_final.pdf)