

What is Cobalamin G and E?

These are rare genetic conditions that occur when the body is unable to process certain amino acids (building blocks of protein) properly. They cause a buildup of the Homocysteine (HCY) and a deficiency in the amino acid, methionine. These are also called the methylcobalamin disorders

What are methylcobalamin disorders?

CblG and CblE are two rare genetic disorders that occur when the body is unable to process vitamin B12 to methylcobalamin which is needed to synthesize methionine, an important building block for the body. CblG and CblE are forms of Homocystinurias (HCU). Children with CblG/CblE have elevated homocysteine and low methionine in their blood.



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 as well as a dangerous buildup 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
- Movement Problems
- Developmental Delay
- Intellectual disability
- Microcephaly
- Cerebral Atrophy
- Neurological disorders
- Blood clots or strokes

How can I help?

Teach your student as you would anyone else. HCY levels can fluctuate. Your student with CblG and CblE may need additional time or attention to keep pace with the classroom.

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 CblG or CblE. Successful HCU management will rely on both parents and school staff com-

Possible Medication Side Effects

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

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 CblG, or CblE 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 CblG and CblE 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 CbIG and CbIE 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 CbIG and CbIE has the same learning opportunities as other students in the classroom.

Accommodations might include:

- ⇒ Preferential seating
- ⇒ Modified textbooks or audio-visual materials
- ⇒ Oral test and visual aids
- ⇒ Low vision Accomodations
- ⇒ Easy access to bathroom and/or nurse
- ⇒ Accommodations for fine and gross motor activities
- ⇒ Assisting with self-help skills
- ⇒ Access to assistive technology

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 CbIG and CbIE .



Additional Services

Because the whole body impact CbIG and CbIE may have, some patients may need additional services.

- ⇒ Vision Itinerant
- ⇒ Speech Therapy
- ⇒ Occupational Therapy
- ⇒ Physical Therapy
- ⇒ Reading Help
- ⇒ Counseling, friendship groups, help with social skills (if necessary)
- ⇒ 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 CbIE and Cbl G, please visit: <https://hcnetworkamerica.org>

Additional resources:

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