Our **Mission**

The HCU Network America strives to inform and provide resources for patients and families, create connections, influence state and federal policy, and support advancement of diagnosis and treatment for HCU and related disorders.

Our **Goals**

To support research that improves diagnosis and treatment including a cure for the disease.

To provide information and resources to better manage the disease.

* HCU treatment and dietary approaches
* General disease management
* Increased access for treatment and supplements
* Research findings and opportunities for clinical trials

To create connections across the community and facilitate sharing of information and best practices through in-person and virtual events and discussions

To assure all patients are diagnosed as early and efficiently as possible to enable access to care and avoid complications

Connect with us

Online: hcunetworkamerica.org

Email: info@hcunetworkamerica.org

Phone: (630) 360-2087

Facebook: HCU Network America

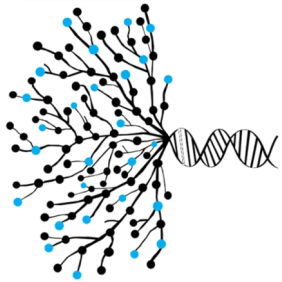
Instagram @HCU_Network_America

Twitter: @HCUAmerica

About us

HCU Network America is a registered 501c3 non profit organization dedicated to helping patients and their families affected by Homocystinuria (HCU), Methylenetetra hydrofolate reductase (MTHFR) and selected cobalamin deficiencies.

HCU Network America



What is HCU?

The Homocystinurias are a group of **inherited metabolic disorders** leading to build up of homocysteine and its metabolites in the blood and urine. **Classical Homocystinuria** (HCU) is believed to be the most common type of these disorders, caused by deficiency in the enzyme known as **cystathionine beta-synthase** (CBS). **Cobalamin** (Cbl) C, D-HCY, E, F, G, X and **Severe MTHFR** are also part of the homocystinuria group. There are other disorders that cause high homocysteine and thus are called homocystinurias, there is little known about them, but we are still learning about them.

How is it Diagnosed?

In America, Classical Homocystinuria is screened for by newborn screening but **screening does not detect all cases**. If not diagnosed by newborn screening, it can take an average of **4.5 years for an accurate diagnosis** of HCU. Early diagnosis and **treatment** can make a **real difference** to patient outcomes. CblC, D-tHCY, and F may be detected by newborn screening in some states, but not all may detect G, E and MTHFR

What are the **symptoms**?

Ocular (Eyes)

* Severe and progressive nearsightedness
* Lens dislocation

Central Nervous System

* Developmental and cognitive deficits
* Seizures
* Clumsiness
* Psychiatric disorders
* Behavioral problems

Skeletal

* Excessive growth of bones
* Protruding or sunken chest
* Highly arched feet

Vascular

* Blood clots
* Strokes

(Mildly affected individuals may present as adults with blood clots as their only problem).

The **non-specific** nature of the signs and symptoms can lead to **under diagnoses**.

A **treatable** disorder

No cure has been identified for Classical or other forms of HCU.

Classical HCU:

There are two forms of treatment:

B6 responsive:
B6-responsive patients respond well to high doses of vitamin B6 and most will also have folic acid supplementation. This may be all the treatment they need.

B6 non-responsive:
B6 non-responsive patients require treatment involving a low protein diet, medical formula to supplement amino acids, betaine, folic acid and sometimes B12.

Cobalamin:

Cobalamin patients should avoid a low protein diet, and should be prescribed Hydroxocobalamin (OHCbl) and betaine, and also may be prescribed a combination of leucovorin, L-Methionine and L-Carnitine.

Severe MTHFR:

Severe MTHFR patients should also avoid a low protein diet. They should be prescribed betaine and may be prescribed a combination of Folonic Acid, 5-Methylfolate , L-Methionine and L-Carnitine.

Milder MTHFR patients are often not treated unless they have high homocysteine levels.

Information provided is for educational purposes only. Please consult your doctor regarding any symptoms you might be experiencing.