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

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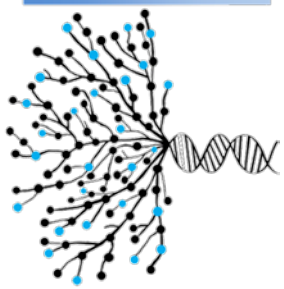
About us

HCU Network America is a registered 501c(3) non-profit organization dedicated to helping patients and their families affected by Homocystinuria (HCU), Methylene-tetrahydrofolate 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 the most common type of these disorders, caused by deficiency in the enzyme known as **cystathionine beta-synthase** (CBS).

How is it Diagnosed?

In America, Homocystinuria is screened by newborn screening but **screening does not detect all cases of Homocystinuria** and is not available in all countries. 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.

The other forms of Homocystinuria are not discussed here.

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 Homocystinuria. Treatment involves a strict low protein diet in conjunction with supplementation. Treatment may vary based on whether the specific defect reults in the patient being responsive to Vitamin B6 (Pyridoxine) or not.

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.

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