HOW IS HCU
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,

4.5 YEARS

Average time for an accurate HCU diagnosis

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

CONNECT WITH US

HCU NETWORK AMERICA

www.hcunetworkamerica.org

info@hcunetworkamerica.org

(630) 360-2087

@HCU Network America

@HCU_Network_America

@HCUNetworkAmerica

@HCUAmerica

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.

What is it?

How is it diagnosed?

What are the symptoms?

How is it treated?

HCU

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, J, 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.

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 underdiagnoses.

HCU IS 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.