CLASSICAL HOMOCYSTINURIA (HCU)

A rare, inherited metabolic disorder requiring lifelong medical nutrition.

Quick Facts

- Classical HCU is screened at birth in all 50 states
- Requires strict dietary therapy for life
- Nitrous oxide, long periods of fasting, can trigger metabolic crises
- Medical foods are medically necessary, not optional
- With proper management and access to care, individuals with Classical HCU can thrive

What Is Classical HCU?

Classical homocystinuria (Classical HCU) is a rare, life-threatening genetic metabolic disorder. Individuals with Classical HCU cannot properly break down the amino acids methionine and homocysteine.

How can it affect the body?

The buildup of homocysteine levels can have dangerous and life-threatening impacts.

<u>High homocysteine levels may cause:</u>

- Severe nearsightedness, dislocation of lenses
- Blood clots/Strokes
- Seizures
- Scoliosis
- Premature osteoporosis
- Cognitive and/or behavioral issues

How is it treated?

Classical homocystinuria is treated with a combination of B6, B12, Folic Acid, Betaine, a low-protein diet, and/or medical formula. Treatment is required for life.

HCU NETWORK AMERICA

About the HCU Network America

HCU Network America is a 501c3 nonprofit dedicated to:

- Supporting individuals and families living with HCU
- Providing education and resources
- Offering webinars, community connections, and family support
- Advocating for newborn screening and access to therapies
- Collaborating with metabolic clinics and rare disease partners

Why HCU Requires Policy Attention

HCU families face challenges that require legislative action:

- Medical foods are essential treatment, not optional nutrition.
- Coverage for medical formula and low-protein foods varies by state and insurance plan.
- Newborn screening saves lives—but states need strong screening systems and reliable follow-up.
- Rare disease research accelerates better treatments and improves long-term outcomes.

Key Challenges Facing HCU Families

- High out-of-pocket costs for medical formula and low-protein foods
- Inconsistent insurance coverage across states
- Variation in newborn screening, follow-up, and resources across states
- Limited rare disease research funding relative to need

How Public Policy Can Help

- Guarantee medical nutrition coverage under private insurance, Medicaid, and CHIP
- Include low-protein medical foods as part of covered treatment
- Strengthen and fund newborn screening programs
- Support rare disease initiatives and research through the National Institutes of Health (NIH) and the Health Resources and Services Administration (HRSA).
- In addition to federal action, advocates can often make faster progress at the state level on newborn screening, medical foods/formula coverage, and rare disease caucus efforts.

Learn More About Newborn Screening & Medical Nutrition

Newborn Screening: https://hcunetworkamerica.org/newborn-screening/

Patients & Providers for Medical Nutrition Equity (Medical Foods/Formula Advocacy): https://nutritionequity.org

HCU NETWORK AMERICA

HCU Network America
https://hcunetworkamerica.org

HCU Data Collection Program
https://rare-x.org/homocystinuria/