INTRACELLULAR COBALAMIN DISORDERS

A rare, inherited metabolic disorder requiring lifelong medical nutrition.

Quick Facts

- Screening is dependent upon the state you are born in.
- Requires high doses of hydroxocobalamin and betaine.
- Illness, fasting, Nitrous oxide, or stress can trigger metabolic crises
- Hydroxocobalamin is medically necessary, not optional
- With proper management and access to care, individuals with intercellular cobalamin disorders can thrive

What is an Intracellular Cobalamin Disorder?

Intracellular cobalamin disorders are a group of rare, life-threatening genetic metabolic disorders that affect the body's ability to process vitamin B12. They are individually named Cobalamin C, D, E, F, G, J, K, and X. Individuals with these disorders cannot properly break down the amino acid 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 vision impairment
- Global developmental delays
- Failure to thrive
- Seizures
- Anemia

How is it treated?

Intracellular cobalamin disorders are treated with a combination of hydroxycobalamin, betaine, and folinic acid. Treatment is required for life.

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 Nutrition is an essential treatment, not optional
- Coverage for hydroxocobalamin 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 hydroxocobalamin
- 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/