SEVERE METHYLENE TETRAHYDROFOLATE REDUCTASE DEFICIENCY

Quick Facts

Screening is dependent upon the state you are born in.

Requires betaine and folinic acid.

Illness, fasting, or Nitrous Oxide can trigger metabolic crises

With proper management and access to care, individuals with Severe MTHFR deficiciency can thrive

This is not the common MTHFR Variants

What is Severe Methylene TetrahydroFolate Reductase Deficiency (Severe MTHFR)

Severe MTHFR deficiency is a rare metabolic disorder that affects the body's ability to process folate, homocysteine, and methionine, leading to serious neurological and vascular complications.

How can it affect the body?

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

High homocysteine levels may cause:

- Global developmental delays
- Failure to thrive
- Low muscle town
- Seizures

How is it treated?

Severe MTHFR is treated with a combination of 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:

- 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

- 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 coverage under private insurance, Medicaid, and CHIP
- 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
https://hcunetworkamerica.org

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