

ABOUT THE FOUNDATION

The **NATIONAL UREA CYCLE DISORDERS FOUNDATION** is a non-profit organization dedicated to the identification, treatment and cure of urea cycle disorders. It is an internationally recognized resource of information and education, and is a supportive network for families, friends and medical professionals.

The goals of our organization are:

- To provide guidance and information to families and others affected by urea cycle disorders.
- To educate medical professionals on the diagnosis and treatment of urea cycle disorders.
- To stimulate and support research which leads to improved treatment for today and a CURE for tomorrow.
- To increase public awareness on the existence of urea cycle disorders so that no child ever goes undiagnosed.
- To educate legislators on the needs of children affected by these rare disorders.

NUCDF is a member of:

Genetic Alliance
National Institutes of Health Rare Disease Clinical Research Network
Urea Cycle Disorders Research Consortium
National Organization for Rare Disorders (NORD)
National Healthy Mothers Healthy Babies Coalition
Preserving the Future of Newborn Screening
SaveBabies.org

Contributions are appreciated and help us achieve our goals and support research.



NATIONAL UREA CYCLE DISORDERS FOUNDATION

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NATIONAL UREA CYCLE DISORDERS FOUNDATION

Dedicated to saving the lives of children and adults with urea cycle disorders

Saving Lives Awareness Bracelet



Did you know that missing one little enzyme in your body could be responsible for brain damage, cerebral palsy, coma and death?

Our only dream is for a cure.

And we can't rest until we find it.

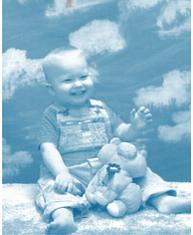
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Every day more infants and children of all ages are being diagnosed with this devastating disorder, and the fight for their lives begins.

What Is A Urea Cycle Disorder?

Urea Cycle Disorders are catastrophic disorders characterized by excessive amounts of ammonia in the blood.

The urea cycle involves a series of biochemical steps that take place in the liver which remove nitrogen, a by-product of protein metabolism, from the blood.



There are six urea cycle enzymes in this cycle. A deficiency in one of these enzymes results in a highly toxic accumulation of ammonia in the bloodstream. Ammonia reaches the brain, where it may cause irreversible brain damage and/or death.

What Are The Symptoms?

Infants may present within 48 hours of birth with irritability, followed by vomiting and increasing lethargy. Seizures, poor muscle tone, respiratory distress, and finally coma occur. The symptoms are caused by rising ammonia levels in the blood.



Toddlers and children with mild to moderate urea cycle enzyme deficiencies commonly go undiagnosed until severe symptoms appear. Early symptoms include hyperactive behavior, screaming or destructive behavior, agitation, irritability, headaches, refusal to eat high-protein foods, and periods of “spaciness” or inattention. Later symptoms include frequent vomiting, lethargy, delirium, seizures and, if untreated, coma.

Children with this disorder are often misdiagnosed with behavioral problems, autism spectrum or ADHD. Childhood episodes may be brought on by viral illness, vaccines, or dehydration.

Adults can also have undiagnosed urea cycle disorders. Stressors on the body such as surgery (i.e., gastric bypass, child-birth), medications like prednisone, and high-protein diets or excessive exercise can unmask an underlying urea cycle disorder. Many of these individuals exhibit psychiatric symptoms (schizophrenia or bipolar), periods of confusion, extreme agitation or disorientation that are mistaken for alcohol or drug intoxication.

How Common Is This Disorder?

Urea Cycle Disorders are categorized as a “rare disease,” but researchers estimate that as many as one in 8,500 children are born with the disorder. Many experts believe the number of affected children is even higher, but the disorder is not properly diagnosed or never diagnosed—it is also believed that many SIDS deaths and autism spectrum cases may actually be due to undiagnosed urea cycle disorders.

As you can imagine, our only dream is for a cure.

And we can't rest until we find it.