what is the Urea Cycle Disorders Consortium

The Urea Cycle Disorders Consortium (UCDC) is a team of doctors, researchers, and patient advocates throughout the US, working together to improve the lives of individuals and families affected by urea cycle disorders through research and education. The consortium provides a way for patients to join doctors and researchers in developing new and better treatments for urea cycle disorders by participating in research studies. The greater the collaboration between doctors and patients, the more we can learn about urea cycle disorders. This important first step is necessary if we are ever to find new and better treatments.

The goals of the UCDC are to:

- Develop better treatments and a deeper scientific understanding of the causes of UCD.
- Understand how UCD can cause brain damage and develop protection against this.
- Conduct clinical trials of promising new drugs for the treatment of UCD.
- Work with the National Urea Cycle Disorders Foundation, the UCD patient advocacy group, to help patients who wish to be involved in research connect with doctors conducting UCD research.
- Construct and maintain resources with significant information for clinicians, researchers, and patients.
- Train physicians to become experts in providing care for and treating those with UCD.

Visit Our Web Site:
www.RareDiseasesNetwork.org/ucdc

UCDC Clinical Centers

Children's Hospital of Philadelphia
Philadelphia, Pennsylvania

Children's National Medical Center
Washington, District of Columbia

Mt. Sinai Hospital
New York, New York

Rainbow Babies and Children's Hospital
Cleveland, Ohio

Texas Children's Hospital
Houston, Texas

UCLA Medical Center
Los Angeles, California

Vanderbilt University Hospital
Nashville, Tennessee

Yale-New Haven Hospital
New Haven, Connecticut

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Much of the body including the organs and muscles is made up of protein. To keep healthy, the body is always making new protein and breaking down old protein that has become damaged or old. The body must also break down the protein we eat into a form it can use. When protein is broken down, a substance called ammonia is formed. Normally, the body deals with this ammonia by combining it with chemicals called enzymes to make a harmless substance called urea.

People with urea cycle disorders (UCDs) are born without one of the enzymes needed to change ammonia into the safer chemical urea in blood and urine. This conversion happens in several steps called the urea cycle. When these enzymes are missing from the body, ammonia can build up and can cause illness, brain damage or death.

what is a Urea Cycle Disorder?

When seen in babies, UCDs can cause the newborn to stop feeding, become sleepy, and often throw up. Sometimes they will also have seizures. If they are not diagnosed and treated quickly, they can die or suffer permanent brain damage. Those with a less severe form of urea cycle disorder may have only mild symptoms and not be diagnosed until later in life. There is no cure for a urea cycle disorder except for liver transplantation, although novel therapies have improved the outcome for many people with UCD.

Because many cases of urea cycle disorders remain undiagnosed and babies born with the disorders often die without a diagnosis, the exact incidence of these disorders is unknown. In addition to studies focusing on improving diagnosis and treatment, the UCDC is conducting research studies to find out the number of people in the population with UCD.

how to contribute

Everyday, research studies are in action, directed at improving our knowledge and treatment of urea cycle disorders. These continuing efforts of researchers seek to improve the quality of life for all who are suffering from these rare disorders, but this work by researchers cannot occur without the vital partnership with patients. By joining our registry, you can become involved in this important effort.

How the Contact Registry works

The UCDC Contact Registry provides a way for patients with UCD to find out about UCD research. Patients, and/or parents of young patients, can register to be contacted about UCDC research by providing basic contact information through a secure form. The secure form is not submitted directly to UCDC researchers, but is managed by the Rare Diseases Clinical Research Network's Data and Technology Coordinating Center. The Data and Technology Coordinating Center will contact patients in the contact registry by email or letter to inform them of clinical research studies performed in our new multi-center Urea Cycle Disorders Consortium. Private information will not be given to researchers. Instead, with the information received about clinical research, patients can decide which research studies they want to learn more about and participate in. The contact registry is free of charge.

help when you need it

The National Urea Cycle Disorders Foundation patient advocacy organization is here to help you. This group is an important partner of the UCDC and is devoted to providing support, information, education and resources to patients and families. This organization promotes research into the cause, treatment, and cure of urea cycle disorders.

National Urea Cycle Disorders Foundation
800-38-NUCDF
www.nucdf.org
info@nucdf.org

Visit Our Web Site:
www.RareDiseasesNetwork.org/ucdc

You can learn more about the Contact Registry at our web site:
www.RareDiseasesNetwork.org/ucdc/takeaction

or by calling Patient Contact Registry Assistance:
866-313-9879

Who can join the Registry?

We encourage patients from the United States and around the globe with a confirmed or suspected diagnosis of one of the eight urea cycle disorders listed below to join the UCDC Contact Registry:

- N-Acetylglutamate Synthase (NAGS) Deficiency
- Carbamyl Phosphate Synthetase I (CPS I) Deficiency
- Ornithine Transcarbamylase (OTC) Deficiency
- Argininosuccinate Synthetase (AS) Deficiency (Citrullinemia)
- Citrin Deficiency (Citrullinemia II)
- Argininosuccinate Lyase (AL) Deficiency (Argininosuccinic Aciduria)
- Arginase (ARG) Deficiency (Hyperargininemia)
- Ornithine Translocase (ORNT) Deficiency (HHH Syndrome)

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