National Urea Cycle Disorders Foundation is the only nonprofit organization in the world solely dedicated to saving and improving the lives of children and adults with urea cycle disorders. NUCDF leads the fight to conquer UCD, and is the driving force behind cutting-edge research. NUCDF serves as a vital resource to medical professionals and a lifeline to affected families all over the world, providing information, support and **Hope**.

Our Vision is a world in which no child or adult suffers or perishes from urea cycle disorders.

Our Mission is to save and improve the lives of all those affected by urea cycle disorders.

The goals of our organization are:

- **Stimulate and support research** which leads to improved quality of life for today and a **CURE** for tomorrow.
- **Provide guidance and information** to families and others affected by UCD.
- **Educate medical professionals** on the identification, diagnosis and treatment of UCD.
- Provide a caring community, **networking families and patients** together for support.
- **Increase public awareness** on the existence of UCD so that no child or adult ever goes undiagnosed.
- **Educate public policy leaders** on the needs of all those affected by these rare disorders.

NUCDF

Our dream is a cure...
and we can't rest until we find it.

To Join the Study
or for more information
contact:

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www.CureUCD.org

Saving lives through
Research, Education, Awareness,
Community and Hope

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UTILITY OF NEXT-GENERATION SEQUENCING DNA TECHNOLOGIES IN DIAGNOSIS OF PROXIMAL AND NOVEL DISORDERS OF UREAGENESIS

New Study for patients with diagnosed or suspected OTC or CPS1 deficiency without DNA mutational confirmation

UTILITY OF NEXT-GENERATION SEQUENCING DNA TECHNOLOGIES IN DIAGNOSIS OF PROXIMAL AND NOVEL DISORDERS OF UREAGENESIS

Background:

Definitive diagnosis of proximal urea cycle defects (NAGS, CPS1, OTC deficiency) is accomplished through DNA mutational analysis. Specifically in OTC deficiency, certain mutational analysis techniques may fail to detect a mutation. Clinicians may decide not to pursue further DNA testing. In these cases, the diagnosis is made based on clinical presentation and interpretation of biochemical findings. As a result, in the absence of confirmation of a specific mutation, it is possible that some OTC patients may carry an inexact diagnosis.

Treatment and management options for the proximal defects may vary. The impact of unconfirmed diagnosis on outcome and quality of life may therefore be substantial. In this study, molecular testing using next-generation sequencing technologies will be provided to eligible patients to assist their clinicians with potential definitive diagnosis. The study will determine the incidence of patients with ambiguous diagnosis, and evaluate the impact of unconfirmed diagnosis on outcome and quality of life from the patient and family perspective.

For more information contact:
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Goals of the study:

- Resolve ambiguous diagnosis for persons currently being treated for proximal urea cycle disorder who are lacking confirmatory molecular diagnosis.
- Improve the understanding of the proportion of patients with proximal urea cycle defects who may carry an ambiguous diagnosis due to lack of access to confirmatory DNA testing.
- Improve the understanding of the burden of ambiguous diagnosis on UCD patients and families.

Participation:

- Participants will have a single blood draw at a local lab for genetic testing for UCD.
- Testing will be shipped to and performed by Baylor Miraca Genetics Laboratories, Houston TX.
- Results will be provided to the physician ordering the testing and the patient will receive a letter explaining the results.

Eligibility:

- Inclusion: Males and females of all ages diagnosed with OTC or CPS1 deficiency via clinical presentation and/or biochemical findings, without DNA mutational analysis or with no mutation found.
- Exclusion: Patients with confirmed molecular diagnosis of any form of UCD; confirmed diagnosis of a distal UCD or transporter defect based on biochemical findings; negative exome or genome sequencing; diagnosis of mitochondrial disorder, fatty acid oxidation disorders, organic acidemias, or other inborn errors of metabolism.