LONGITUDINAL STUDY OF UREA CYCLE DISORDERS

Summary / Zusammenfassung
The objective of this international multicenter study is to conduct a longitudinal multidisciplinary investigation of the natural history, morbidity, and mortality in people with urea cycle disorders. Urea cycle disorders are a group of rare inborn errors of metabolism that commonly present in childhood with episodes of vomiting, lethargy and coma. Symptoms are the result of an accumulation of ammonia, a toxic product of protein degradation, which is not adequately metabolized in the liver of affected individuals due to an enzyme deficiency present from birth. The risk of death or severe disability from these disorders approaches 50% and current therapy is considered inadequate. The purpose of this study is to perform a long-term follow-up of a large group of patients with the various urea cycle disorders, including assessment of growth, cognitive function and biochemical status over time. Survival and cognitive outcome of current treatment forms will be evaluated and identification of biochemical changes (biomarkers) that may predict future metabolic imbalances will be sought so that they can be corrected before clinical symptoms develop. The overall goal of the study is to improve treatment and outcome of this devastating group of disorders.

The study is carried out within the framework of the Urea Cycle Disorders Consortium (UCDC), which is part of the Rare Diseases Clinical Research Network (RDCRN) established by the National Institutes of Health (NIH) in the USA.

Publications / Publikationen

Keywords / Suchbegriffe
urea cycle disorders, natural history, ornithine transcarbamylase, carbamoylphosphate synthase, N-acetylglutamate synthase, argininosuccinate synthase, argininosuccinate lyase, arginase, ORNT1

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**In Collaboration with / In Zusammenarbeit mit**

The international study chairs: Mark Batshaw, MD Marshall Summar MD and Mendel Tuchman, MD and the participating academic institutions:

Children’s National Medical Center (CNMC), Washington, DC
Children's Hospital of Philadelphia (CHOP)
Baylor Medical Center, Houston, Texas
Vanderbilt University Medical Center, Nashville, Tennessee
University of California Los Angeles (UCLA)
Mt. Sinai School of Medicine, New York, New York
Rainbow Babies and Children’s Hospital, Cleveland, Ohio
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The Children’s Hospital, Aurora, Colorado,
Children’s Hospital, Boston, Massachusetts
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**Duration of Project / Projektdauer**

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