

# Urea Cycle Disorders Consortium

## UCD Research Studies Recruiting Participants

If you are interested in our studies, please visit the [UCDC website](#) to learn about enrollment and participating sites near you.

### **5101: Longitudinal Study of UCDs**

Long-term observation of the impact of UCDs on growth and development, including physical, neurological and cognitive functioning. Studies the relationship between health indicators and disease severity and outcomes for those using different UCD therapies, including liver transplant.

### **5111: Carbaglu® Surveillance Protocol**

A post-FDA approval surveillance of carglumic acid (Carbaglu) to obtain long-term clinical safety information. In addition to natural history data collected through protocol 5101, this study collects data on adverse reactions and pregnancy outcomes in those taking Carbaglu for treatment of N-acetylglutamate synthase (NAGS) deficiency.

### **5113: Biomarkers of Neurological Injury and Recovery in Urea Cycle Disorders**

Studies how UCDs affect thinking, body chemistry and brain structure using magnetic resonance imaging (MRI) and behavioral testing. Individuals who do not have UCDs participate to compare healthy brain function to brain function impacted by UCDs.

### **5119: Electrographic Seizures in Hyperammonemia**

Investigates the presence of electrographic seizures during hyperammonemic events and if seizure intensity is connected to abnormal MRI findings, the development of epilepsy and neurological damage.

### **5120: Noninvasive Biomarkers of Hepatic Fibrosis in UCDs**

Determines if non-invasive procedures—procedures that do not require instruments to enter the body— such as ultrasound and laboratory testing, can indicate increased risk for liver stiffness.

### **5121: Neurodevelopmental Assessments: Comparison of Standard (Traditional) Neuropsychological Battery and NIH Toolbox**

Compares the NIH Toolbox, a computerized system of neuropsychological testing created by the National Institutes of Health (NIH), to standard neuropsychological testing currently used in UCDC studies. Also assesses how well the Toolbox is received by participants and testing administrators. (Supplement to 5101)

### **5122: Hepatic Histopathology in UCDs**

Analyses past and present data to estimate the proportion of UCD individuals with liver disease, and compares the presence of liver disease across UCD subtypes and treatments, such as liver transplantation.

### **5123: Novel Food Photography Method to Measure Dietary Intake in Patients with UCDs**

Participants keep a visual food diary using a photography app. The accuracy and ease of use of the calorie and protein intake calculated by the app is compared to manual calculation.