# Longitudinal Study of Urea Cycle Disorders

NCT00237315

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| **Status** | RECRUITING |
| **Sponsor** | Andrea Gropman |
| **Enrollment** | 1,500 participants |

## Key Eligibility Criteria

### Inclusion (9)

- Diagnosis of NAGS deficiency, defined as the detection of a pathogenic mutation, and/or decreased (less than 20 % of control) NAGS enzyme activity in liver ,and/or hyperammonemia and first degree relative meets at least one of the criteria for NAGS deficiency
- Diagnosis of CPS I deficiency, defined as decreased (less than 20 % of control) CPS I enzyme activity in liver, and/or an identified pathogenic mutation, and/or hyperammonemia and first degree relative meets at least one of the criteria for CPS I deficiency
- Diagnosis of OTC deficiency, defined as the identification of a pathogenic mutation, and/or less than 20% of control of OTC activity in the liver, and/or elevated urinary orotate (greater than 20 uM/mM) in a random urine sample or after allopurinol challenge test, and/or hyperammonemia and first degree relative meets at least one of the criteria for OTC deficiency
- Diagnosis of AS deficiency (Citrullinemia), defined as a greater than or equal to 10-fold elevation of citrulline in plasma, and/or decreased AS enzyme activity in cultured skin fibroblasts or other appropriate tissue, and/or identification of a pathogenic mutation in the AS gene, and/or hyperammonemia and first degree relative meets at least one of the criteria for AS Deficiency
- Diagnosis of AL deficiency (Argininosuccinic Aciduria, ASA), defined as the presence of argininosuccinic acid in the blood or urine, and/or decreased AL enzyme activity in cultured skin fibroblasts or other appropriate tissue, and/or identification of a pathogenic mutation in the AL gene, and/or hyperammonemia and first degree relative meets at least one of the criteria for AL Deficiency

... and 4 more (see full listing online)

### Exclusion (2)

- Hyperammonemia caused by an organic academia, lysinuric protein intolerance, mitochondrial disorder, congenital lactic academia, fatty acid oxidation defects, or primary liver disease
- Rare and unrelated comorbidities (e.g., Down's syndrome, intraventricular hemorrhage in the newborn period, and extreme prematurity)

## Locations (15 total)

University of California, Los Angeles, Los Angeles, California, United States
Stanford University Medical Center, Stanford, California, United States
Children's Hospital Colorado, Aurora, Colorado, United States
... and 12 more locations