## **Condition:** Urea cycle disorders

---

### Inheritance:

Autosomal recessive or X-linked recessive (OTC deficiency).

### Genetic etiology:

Genetically heterogeneous, due to mutation in genes encoding enzymes of urea cycle.

### Frequency:

Approximately 1/30,000 births collectively.

### Clinical features:

The urea cycle functions to convert ammonia to urea. Deficiency of enzymes in the pathway lead to toxic buildup of ammonia, which presents with acute encephalopathy, leading to coma and death if not treated.

### Management:

Acute episodes may require dialysis to reduce ammonia concentrations. Chronic treatment with sodium benzoate and sodium phenylacetate provide alternative pathways for ammonia excretion.

### Genetic counseling:

Depends on specific genetic cause; most are autosomal recessive, except for ornithine transcarbamylase deficiency, which is X-linked. Molecular genetic testing is available.

*Human Genetics and Genomics*, Fourth Edition. Bruce R. Korf and Mira B. Irons.