Condition: Organic acidemia (proprionic academia; methylmalonic academia; isovaleric academia; glutaricacidemia I and others)

Inheritance:

Autosomal recessive.

Genetic etiology:

Due to mutation in various genes encoding components of enzymes involved in organic acid metabolism.

Frequency:

Collectively approximately 1/1,000.

Clinical features:

Presentation shortly after birth with neonatal encephalopathy associated with metabolic acidosis, ketones in the urine, and hyperammonemia. Metabolic crises may recur even after treatment is instituted due to intercurrent illness.

Management:

In general, the mainstay of treatment is careful monitoring of specific amino acid or protein intake and prevention of a catabolic state. In some cases, specific coenzymes can be administered. Liver transplantation has been used successfully in some cases.

Genetic counseling:

Based on autosomal recessive inheritance; disorders may be detected by expanded newborn screening using tandem mass spectrometry. Genetic testing available for most disorders.

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