Condition: Tyrosinemia

Inheritance:

Autosomal recessive.

Genetic etiology:

Mutation in *FAH* gene encoding fumarylacetoacetase.

Frequency:

Approximately 1/100,000 births.

Clinical features:

Tyrosinemia presents in infants or young child with liver failure and renal dysfunction, and eventual development of hepatocellular carcinoma. Untreated children develop neurological episodes of altered mental status, abdominal pain, and respiratory failure. Most do not survive untreated past 10 years.

Management:

Administration of nitisinone, which inhibits tyrosine degradation, avoiding toxic buildup of fumarylacetoacetate and succinylacetone; low tyrosine diet.

Genetic counseling:

Based on autosomal recessive inheritance; molecular genetic testing available.

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