Condition: Galactosemia

Inheritance:

Autosomal recessive.

Genetic etiology:

Mutation in *GALT,* encoding galactose-1-phosphate uridyl transferase.

Frequency:

Approximately 1/30,000.

Clinical features:

Galactosemia presents in infancy with failure to thrive, liver failure, bleeding, and *E. coli* sepsis. Severe mental retardation ensues if treatment is not instituted. Galactosemia is included in newborn screening in most areas. Longterm complications, even in setting of dietary treatment, can include cognitive impairment, cataracts, and premature ovarian failure.

Management:

Dietary restriction of galactose.

Genetic counseling:

Based on autosomal recessive inheritance; molecular genetic testing is available.

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