deficiencies. The most common type of galactosemia (classic galactosemia) results from a deficiency of a hepatic enzyme, galactose 1-phosphate uridyltransferase (GALT), and affects approximately 1 in 50,000 births. The other two varieties of galactosemia involve deficiencies in the enzymes galactokinase (GALK) and galactose 4′-epimerase (GALE); these are extremely rare disorders. All three enzymes (GALT, GALK, and GALE) are involved in the conversion of galactose into glucose.

As galactose accumulates in the blood, several organs are affected. Hepatic dysfunction leads to cirrhosis, resulting in jaundice in the infant by the second week of life. The spleen subsequently becomes enlarged as a result of portal hypertension. Cataracts are usually recognizable by 1 or 2 months of age; cerebral damage, manifested by the symptoms of lethargy and hypotonia, is evident soon afterward. Infants with galactosemia appear normal at birth, but within a few days of ingesting milk (which has a high lactose content), they begin to experience vomiting and diarrhea, leading to weight loss. *E. coli* sepsis is also a common presenting clinical sign. Death during the first month of life is frequent in untreated infants. Occasionally classic galactosemia is seen with milder, chronic manifestations, such as growth failure, feeding difficulty, and developmental delay. This presentation is more frequent among African-American children with galactosemia (Kaye, Committee on Genetics, Accurso, et al, 2006).

Diagnostic Evaluation

Diagnosis is made on the basis of the infant's history, physical examination, galactosuria, increased levels of galactose in the blood, and decreased levels of GALT activity in erythrocytes. The infant may display characteristics of malnutrition; hypoglycemia, jaundice, hepatosplenomegaly, sepsis, cataracts, and decreased muscle tone (Bosch, 2006). Newborn screening for this disease is required in most states. Heterozygotes can also be identified because heterozygotic individuals have significantly lower levels of the essential enzyme.

Therapeutic Management

During infancy, treatment consists of eliminating all milk and