Galactosemia

What is Galactosemia?

It is an inborn error in carbohydrates due to a deficiency in one of the enzymes (galactose-1-phosphate uridyl transferase) that is involved in the breakdown of simple sugar galactose. This enzyme is called galactose-1-phosphate uridyl transferase; Galactose is primarily a part of a larger sugar called lactose, which is found in all dairy products and many baby formulas. The signs and symptoms of galactosemia result from an inability to use galactose to produce energy.

What causes the disease?

Mutations in the *GALT* gene are a potential cause of the galactose-1-phosphate uridyl transferase enzyme deficiency.

What are the clinical features of the disease?

The affected babies are usually normal at birth, however, at their first few weeks of life, after drinking milk that contains lactose, they start presenting symptoms such as vomiting, diarrhea, weight loss, failure to gain weight, poor feeding, jaundice, lethargy, hypoglycemia, liver damage, cataract, bleeding, and E. coli sepsis. Even with early treatment, however, children with galactosemia are at an increased risk for developmental delays, speech problems (verbal dyspraxia), abnormalities of motor function, and osteoporosis. In females, premature ovarian failure is possible.

How is the diagnosis confirmed?

The diagnosis of Galactosemia is established by measuring the amount of galactose, galactose-1-phosphate, and enzymatic activity in the blood sample and confirmed by DNA molecular testing of the *GALT* gene.

What is the treatment of the disease?

A galactose-restricted diet is effective in preventing many of the complications of galactosemia, including the liver and kidney problems. It may also reduce the risk for developmental delays. A Biochemical genetics specialist and a Metabolic Genetics dietitian should coordinate the treatment.