Galactosemia

**Alternative Names**  
Galactose-1-Phosphate Uridylyltransferase Deficiency  
Galt Deficiency  
Galactosemia, Classic

**WHO International Classification of Diseases**  
Endocrine, nutritional and metabolic diseases

**OMIM Number**  
230400

**Mode of Inheritance**  
Autosomal recessive

**Gene Map Locus**  
9p13

**Description**  
Galactosemia is a condition wherein the body is unable to metabolize the sugar galactose. Galactose is one of the sugars present in the disaccharide lactose of milk, and therefore, is present in all dairy products. Galactose is normally converted to glucose and digested by the body. However, in the case of galactosemia, this conversion is unable to take place, leading to the accumulation of galactose and its derivatives, such as galactose-1 phosphate and galactitol, in the blood stream. The accumulation of such toxic compounds leads to hepatomegaly, renal failure, cataracts, and brain damage. Continued feeding of milk to galactosemic infants, without recognizing the disease, can lead to serious consequences, including liver cirrhosis, jaundice, partial blindness, and mental retardation. Classical galactosemia is only one of the three different forms of galactosemias observed; all caused by mutations in different genes.

Galactosemia is a comparatively rare disorder, occurring approximately 1 in every 30,000 live births. Diagnostic tests are now available for newborns that can detect the amount of galactose-1 phosphate and the activity of the galactose metabolizing enzymes in the blood. Post-diagnosis, treatment consists of avoiding all milk and milk containing foods. Instead, soy, meat based, and other lactose free formulas can be used.

**Molecular Genetics**  
The classical form of galactosemia is caused by the deficiency of the enzyme galactose-1 phosphate uridyl transferase. This protein is coded for by the gene GALT, located on chromosome 9p13. The GALT enzyme catalyzes the conversion of galactose-1 phosphate to UDP galactose, an important step in the metabolism of the sugar. Two variants of the type I galactosemia are recognized. In the classic variant, the gene is mutated in such a way that almost no enzyme activity is left. However, the Duarte variant is a much milder form, with at least 5 to 20 percent of the enzyme activity remaining.

**Epidemiology in the Arab World**

**Algeria**  
Gillot et al. (1957) described what they described as the first case of congenital galactosemia in Algeria. No further details could be obtained.

**Egypt**  
Badawy et al. (2003) undertook a clinical and nutritional study on two infants diagnosed with classical galactosemia. Full clinical history was taken for the patients, and pedigree analysis was performed along with clinical examination, anthropometric measurements, and developmental evaluation using the Bayley scale of infant development. The GALT enzyme levels were also assayed. Both patients showed delayed developmental milestones, jaundice, hepatomegaly, increased liver functions, cataract and mental retardation. Five
formulas consisting of various combinations of whole wheat, rice, sweet potato, peanut, sesame seeds, corn starch, and sucrose, were prepared, and administered to the patients along with calcium supplementation. Examination after this treatment showed improvements in the biochemical, developmental evaluation, and anthropometric features of both the patients. Badawy et al. (2003) concluded that this diet had a positive impact on the course of the disease, and that the diet should be made on a wider scale to treat Egyptian patients with galactosemia.

**United Arab Emirates**
The Centre for Arab Genomic Studies Working Group conducted a retrospective study for organic acid disorders described at AlWasl Hospital in Dubai between 1995 and 2004. Only one case of galactosemia was observed in a 4-days-old girl. No further details could be obtained.

**References**

**Contributors**
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