

Galactosemia: Updates for the Primary Care Provider

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Galactosemia is an inborn error of metabolism that leads to elevated levels of galactose in circulation. There are three types of galactosemia which vary in severity, depending on what enzyme they affect and to what extent. This paper concentrates on classic galactosemia which is the most severe type and which is an autosomal recessive disorder that results in nearly undetectable galactose-1-phosphate uridylyltransferase, an enzyme that is used to metabolize galactose, activity. This rare metabolic disorder is estimated to affect 1/50,000 infants born in the US. In the neonatal period, classic galactosemia can cause vomiting, jaundice, and liver failure. Upon rapid implementation of a lactose and galactose restricted diet, the acute signs and symptoms of the neonatal period resolve. Long-term management of galactosemia involves close monitoring to detect potential chronic complications such as central nervous system deficits, reproductive struggles, bone health abnormalities, cataracts, and psychosocial difficulties. Current guidelines for the monitoring of these complications are outlined by the Galactosemia Network, or GalNet, a private multinational organization. GalNet also outlines the suggested long-term dietary guidelines recommended for patients with galactosemia. These dietary guidelines focus on limiting the amount of consumed lactose and galactose. New research shows that strict dietary management may not be as helpful at preventing complications as once thought, so future guidelines may allow for more lenient dietary restrictions.