

Disease Name:

GALACTOSEMIA VARIANT

DUARTE, LOS ANGELES, NEGRO VARIANTS; GALACTOSE-1-PHOSPHATE URIDYL TRANSFERASE (GALT) VARIANT

Classification: Carbohydrate disorder

Genetic Information:

Inheritance: Autosomal Recessive

Population Incidence: 1:6,000

Ethnic Incidence: Caucasian

Gene & Location: 9p13; galactose-1-phosphate uridylyltransferase (GALT) gene

Common Mutation: N314D (Duarte), N314D/L218L (LA), S135L (Negro)

OMIM # 230400

Disease Information:

Symptom Onset: Most infants with variant galactosemia will remain asymptomatic. If symptoms develop, they will be in the early months of life.

Symptoms: Prolonged jaundice, liver disease and cataracts may be present, but most are asymptomatic. Variant galactosemics are not at risk for sepsis or metabolic decompensation. Variant forms of galactosemia confer reduced, but not absent, GALT activity ranging from 5-25% of normal.

Physical Findings: None

Treatment: Galactose restriction for the first year of life if the infant is symptomatic, otherwise no treatment necessary.

Natural History without treatment: A few infants with variant galactosemia will be symptomatic in the newborn period or may have elevated galactose-1-phosphate in red cells, although this will resolve and disappear by 6-12 months of age. Most remain asymptomatic and normal. No evidence for the long-term problems associated with classical galactosemia.

Natural History with treatment: Treatment with lactose restriction is controversial. Treatment if necessary is only needed for a year. Growth and development remain normal. It is very important to counsel families and patients carefully so they understand the benign nature of this mutation and do not assume they have classical disease. (link to parent handout)

Metabolic Information:

Missing Enzyme & Location: Galactose-1-phosphate uridylyltransferase; erythrocytes, liver, skin

Newborn Screening Profile: Abnormal GALT enzyme; normal to mild elevation of galactose and galactose-1-phosphate (generally less than 30mg/dl)

Prenatal testing: NA

Miscellaneous Information: Diagnosis is confirmed through GALT enzyme analysis in red blood cells with or without an elevated galactose-1-phosphate in red cells.

Prepared for the NW Regional Newborn Screening Program by Judi Tuerck RN MS

References:

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7. Kelly S, Desjardins L, Khera SA. A Duarte variant with clinical signs. *J. Med. Genet.* 9: 129-131, 1972
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