

Disease Name:

GALACTOSEMIA

GALACTOSE-1-PHOSPHATE URIDYLTRANSFERASE DEFICIENCY, GALT DEFICIENCY, CLASSICAL GALACTOSEMIA

Classification: Carbohydrate disorder

Genetic Information:

Inheritance: Autosomal recessive
Population Incidence: 1:60,000
Ethnic Incidence: Caucasian (1:47,000)
Gene & Location: 9p13; galactose-1-phosphate uridytransferase gene
Common Mutation: Q188R, K285N (70% Caucasian)
>130 mutations known
OMIM # 230400

Disease Information:

Symptom Onset: Within 1-2 days of milk ingestion
Symptoms: Hepatomegaly, jaundice, hypoglycemia, failure to thrive, vomiting, coagulopathy which may lead to retinal hemorrhage. If the diagnosis is not made, 30% will die from sepsis (E. coli). Survivors develop mental retardation, cataracts, verbal and motor dyspraxia, ovarian failure occurs in 80% of females usually before the third decade.
Physical Findings: No specific dysmorphisms.
Treatment: Lactose/galactose free diet; soy formula.
Natural History without treatment: Mental retardation, cataracts, liver disease, ovarian failure in females and verbal/motor dyspraxia.
Natural History with treatment: Cataracts and liver disease can be prevented with galactose restriction; IQ's as a group are about 10 points lower than sibs, but individuals range from severe retardation to normal. Ovarian failure occurs in 80% of females; approximately 50% of all patients have varying degrees of motor and speech dyspraxia despite lactose restriction. Dyspraxia directed speech therapy is helpful.

Metabolic Information:

Missing Enzyme & Location: Galactose-1-phosphate uridytransferase; erythrocytes, liver, skin.
Newborn Screening profile: Elevated galactose (30mg/dl); abnormal GALT enzyme. It is not unusual for tyrosine and methionine to be elevated secondary to liver disease.
Prenatal testing: Chorionic villus, amniocentesis, DNA mutation analysis.

**Miscellaneous
Information:**

Diagnosis is confirmed through GALT enzyme analysis in red blood cells and an elevated galactose-1-phosphate in red cells. Urine Clinitest (not Clinistix) will be positive for galactose.

Prepared for the NW Regional Newborn Screening Program by Judi Tuerck, RN MS, Oregon Health & Science University

References:

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