Disease Name	**Galactosemia**
Acronym	GALT
Disease Classification	Disorder of carbohydrate metabolism
Variants	Yes
Variant name	Duarte galactosemia
Symptom onset	Infancy
Symptoms	The affected infant may appear normal at birth. Within a few days to two weeks after initiating milk feedings, the infant develops vomiting, diarrhea, lethargy, jaundice, and liver damage. Untreated, the disorder may result in death, frequently associated with E. coli septicemia. Infants surviving the above symptoms may evidence developmental delays, hepatomegaly, Fanconi's syndrome, growth failure and cataracts.
Natural history without treatment	If not detected immediately, it results in liver disease, cataracts, mental delays, and even death. Death can occur as early as one to two weeks of age from severe escherichia (E. coli) bacterial infections. E. coli infections are common in untreated galactosemic infants. The American Liver Foundation recommends that all infants who develop jaundice be considered for galactosemia.
Natural history with treatment	As Galactosemic children get older they may encounter delays in speech and females may suffer from ovarian failure. Nevertheless, children who are diagnosed early have very good long-term outlooks and will lead normal, healthy lives.
Treatment	Treatment for galactosemia is the elimination of galactose and lactose from the diet throughout life. Infants are places on soy formula.
Physical phenotype	No abnormalities present at birth.
Inheritance	Autosomal recessive
General population incidence	1:65,000 live births
OMIM Link	http://www.ncbi.nlm.nih.gov/omim/606999
Genetests Link	www.geneclinics.org
Support Group	Parents of Galactosemic Children, Inc. http://www.galactosemia.org Children's Liver Alliance http://www.liverkids.org.au Children Living with Inherited Metabolic Diseases http://www.climb.org.uk/

Newborn Screening ACT Sheet
[Absent/Reduced Galactose-1-Phosphate Uridyltransferase (GALT)]
Classical Galactosemia

Differential Diagnosis: Galactosemia (galactose-1-phosphate uridyltransferase [GALT] deficiency); GALT heterozygotes; GALT variants; artifactual reductions due to enzyme inactivation by high temperature and/or humidity.

Condition Description: In galactosemia, GALT deficiency results in accumulation of galactose-1-phosphate (Gal-1-P) and galactose, causing multi-organ disease.

YOU SHOULD TAKE THE FOLLOWING ACTIONS IMMEDIATELY:

- Contact family to inform them of the newborn screening result, ascertain clinical status, arrange immediate clinical evaluation, stop breast or cow's milk and initiate non-lactose feeding (powder-based soy formula).
- Consult with metabolic specialist; refer if considered appropriate.
- Evaluate the infant (jaundice, poor feeding, vomiting, lethargy, bulging fontanel, and bleeding) and arrange diagnostic testing as directed by metabolic specialist.
- Emergency treatment as recommended by metabolic specialist. If baby is sick, stop cow's milk and initiate non-lactose feedings.
- Educate family about importance of diet change.
- Report findings to newborn screening program.

Diagnostic Evaluation: Quantification of erythrocyte galactose-1-phosphate (Gal-1-P) and GALT. Classical galactosemia shows <1% GALT activity and markedly increased Gal-1-P. Transfusions in infant can invalidate the results of erythrocyte enzyme assays. Enzyme variants may be distinguished by GALT electrophoresis or mutation analysis.

Clinical Considerations: Classical galactosemia presents in the first few days of life and may be fatal without treatment. Signs include poor feeding, vomiting, jaundice and, sometimes, lethargy and/or bleeding. Neonatal *E. coli* sepsis can occur and is often FATAL. Treatment is withdrawal of milk and, if symptomatic, emergency measures.

Additional Information:

Gene Reviews
Genetics Home Reference

Referral (local, state, regional and national):

Testing
Clinical Services