Primary or Secondary Hypergalactosemia (Galactose Elevated)

Increased galactose
First or second screen

Quantitative RBC GALT assay

RBC GALT normal

Urine reducing substance assay

Positive

Galactokinase assay

Reduced

Galactokinase deficiency

Normal

Liver disease

Hepatic A-V malformation

Citrin deficiency

Other inborn error of metabolism

RBC GALT absent/reduced

See GALT deficiency algorithm; determine whether life-threatening form of galactosemia

Negative

Epimerase assay

Positive

Epimerase deficiency

False positive

Negative

Reduced

Galactokinase deficiency

Liver disease

Hepatic A-V malformation

Citrin deficiency

Other inborn error of metabolism

Abbreviations/Key
- GALT = Galactose-1-phosphate uridyltransferase
- A-V = Arteriovenous
- RBC = Red blood cell

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

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