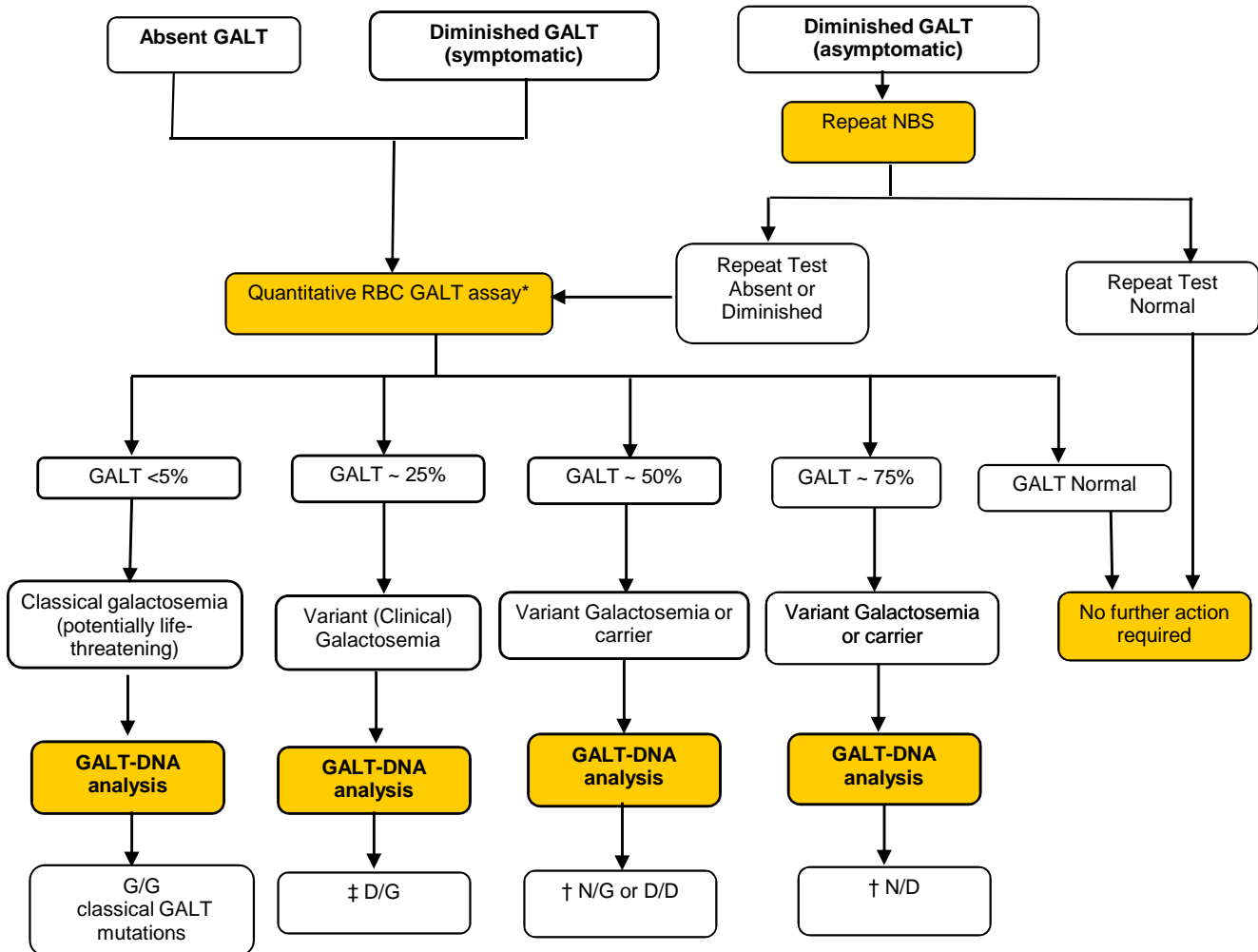


ABSENT OR REDUCED GALACTOSE-1-PHOSPHATE URIDYLTRANSFERASE (GALT)



Abbreviations/Key

GALT = Galactose-1-phosphate uridylyl transferase

NBS = Newborn Screening

RBC = Red blood cell

‡ = Clinically significant variant type

† = Benign variant type

* = Transfusions can invalidate results of RBC enzyme assays

D/D = Duarte homozygote, normal phenotype (~50% GALT activity)

D/G = Duarte/classical galactosemia compound heterozygote (~25% GALT activity)

G/G = classical galactosemia homozygote (<1% GALT activity)

N/D = normal/Duarte carrier (75% GALT activity)

N/G = normal/classical galactosemia carrier (~50% GALT activity)

Action steps are shown in gold (shaded) boxes; results are in plain boxes.

DISCLAIMER: These algorithms and guidelines were adapted from the American College of Medical Genetics algorithm sheets. They are designed primarily as an educational resource for physicians to help them provide quality medical services. Adherence to these standards and guidelines does not necessarily ensure a successful medical outcome. These standards and guidelines should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonable directed to obtaining the same results. In determining the propriety of any specific procedure or test, the healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from these standards and guidelines