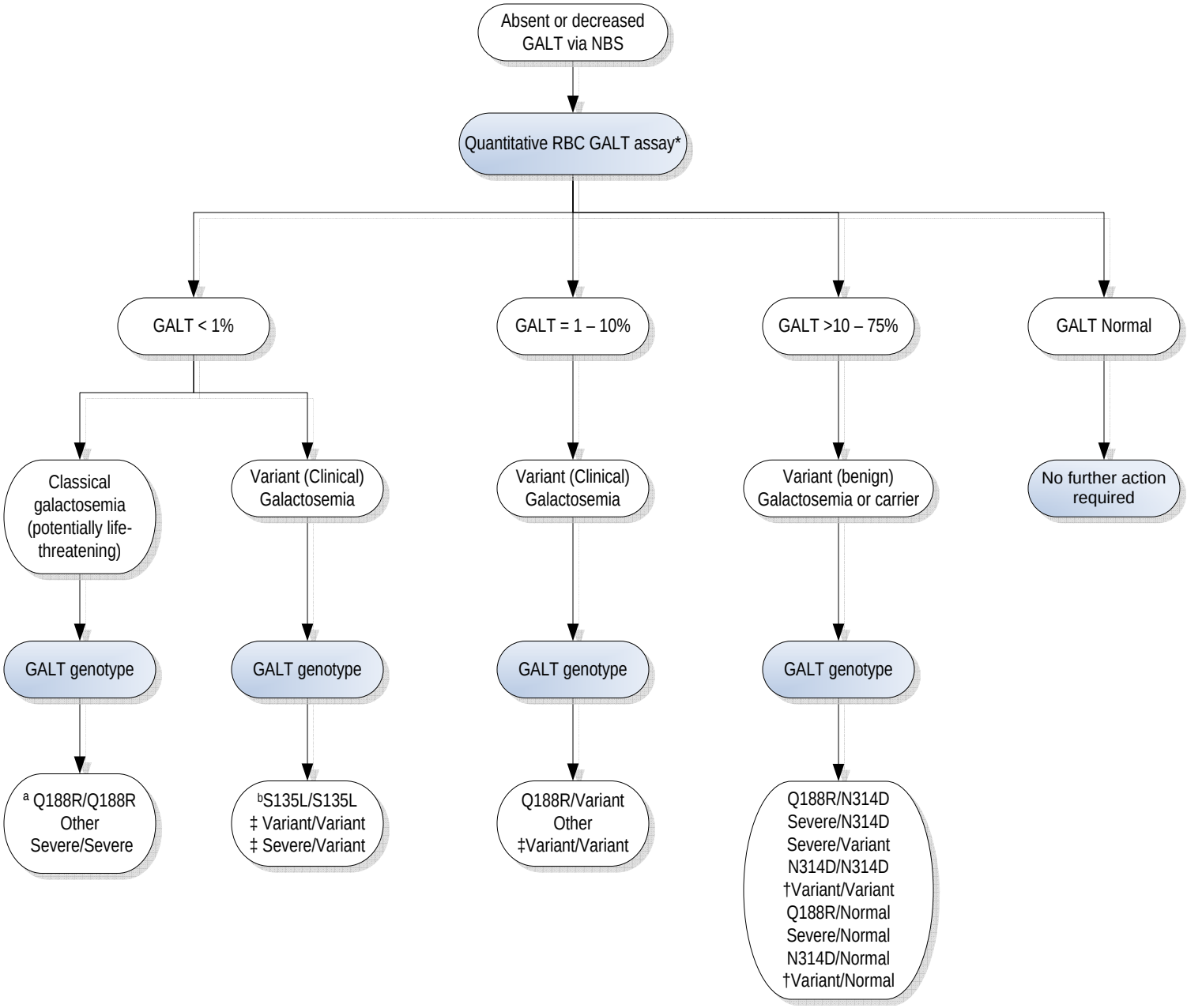
Galactosemia (GALT Deficiency)



Actions are shown in shaded boxes; results are in the unshaded boxes.

Abbreviations/Key

NBS = Newborn screening
GALT = Galactose-1-phosphate uridyl transferase
RBC = Red blood cell
a = Abnormal 120 min. [13C] galactose breath test
b = Normal 120 min. [13C] galactose breath test and 10% GALT in liver

‡ = Clinically significant variant type
† = Benign variant type
* = Transfusions can invalidate results of RBC enzyme assays

Disclaimer: These standards and guidelines are designed primarily as an educational resource for physicians to help them provide quality clinical 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 reasonably 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 record the rationale for any significant deviation from these standards and guidelines.

© American College of Medical Genetics, 2006

(Funded in part through MCHB/HRSA/HHS grant #U22MC03957)