Abbreviations/Key

GALT = Galactose-1-phosphate uridyl 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.