Newborn Screen Follow-up for Glucose-6-Phosphate Dehydrogenase (G-6-PD) Deficiency

Decreased G-6-PD

G6PD / Glucose-6-Phosphate Dehydrogenase (G-6-PD), Quantitative, Erythrocytes*

G6PD activity – Deficient

G6PD deficiency confirmed

Referral to Genetics Specialist

optional

G6PDB / Glucose-6-Phosphate Dehydrogenase (G6PD) Full Gene Sequencing***

G6PD activity – Normal**

Not G6PD deficiency

Assess clinically routine labs: Bilirubin, complete blood count with differential, blood smear, reticulocyte count*

* Reticulocytosis, which can occur to compensate for anemia, and can result in normal G6PD test result in females who are carriers

** In infants with hyperbilirubinemia, consider repeat G6PD enzyme testing at 3 mos of age; hemolysis can result in falsely elevated G6PD levels

*** G6PDB deletion/duplication testing should be considered if sequencing is not informative