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