

THIS IS NOT A TEST REQUEST FORM. Please complete and submit with the test request form or electronic packing list.

G6PD DEFICIENCY TESTING PATIENT HISTORY FORM

Patient Name: _____ **Date of Birth:** _____

Sex Assigned at Birth: Female Male Intersex **Gender Identity (optional):** Female Male _____

Ordering Provider: _____ **Provider's Phone:** _____

Practice Specialty: _____ **Provider's Fax:** _____

Genetic Counselor: _____ **Counselor's Phone:** _____

Patient's Ethnicity/Ancestry (check all that apply)

African American/Black Asian Hispanic White Other: _____

List country of origin (if known): _____

Did the patient have an abnormal newborn screen for G6PD deficiency?..... No Yes

If yes, describe result: _____

Does the patient have symptoms? No Yes (check all that apply and describe)

- Acute hemolytic anemia after exposure to oxidative stress (infection/certain medications/fava beans) Jaundice or hyperbilirubinemia
- Chronic non-spherocytic hemolytic anemia Other Symptom(s): _____

Has the patient undergone previous DNA testing for G6PD deficiency? No Yes Unknown

If yes, describe the test(s) and results: _____

Laboratory Findings

G6PD quantitative enzyme level: Normal Abnormal Unknown Not Performed

Is there any relevant family history?..... No Yes Unknown

If yes, attach a pedigree or specify the relative's relationship to the patient. List their symptoms and age at diagnosis:

Has DNA testing been performed for the family member(s)? No Yes Unknown

If yes, attach a copy of the relative's DNA laboratory result (REQUIRED for familial mutation testing).

Check the test you intend to order.

- 0080135 Glucose-6-Phosphate Dehydrogenase:** Quantitative enzymatic test. Preferred initial test to confirm a diagnosis of *H7QE* deficiency. Clinical sensitivity ~99%; reduced in neonates, heterozygous females, after blood transfusion, or during hemolytic crises.
- 3004457 Glucose-6-Phosphate Dehydrogenase Deficiency (G6PD) Sequencing:** Preferred test for detection of *H7QE* variants in females or any individual with reduced G6PD enzyme activity. Clinical sensitivity ~98%.
- 0051684 Glucose-6-Phosphate Dehydrogenase (G6PD) 2 Mutations:** Detects *H7QE* A- and A+ variants common in individuals of sub-Saharan African descent.
- 2001961 Familial Mutation, Targeted Sequencing:** Tests for sequence variant(s) previously identified in the family. Contact an ARUP genetic counselor at 800-242-2787 ext 2141 prior to test submission to discuss requirements.

Master Label

For questions, contact an ARUP genetic counselor at 800-242-2787 ext. 2141.