

FILE DEFINITION TEST CHANGE

Notification Date: July 15, 2025 Effective Date: August 14, 2025

Glucose-6-Phosphate Dehydrogenase (*G6PD*) Full Gene Sequencing, Varies

Test ID: G6PDZ

Explanation: On the effective date, the Test Algorithm will be updated to include a reflex test when necessary. Additionally, the Specimen Required and Days Performed sections will be updated as indicated below.

Current Reflex Tests	
None	

New Ref	ew Reflex Tests			
Test ID	Reporting Name	Available Separately	Always Performed	
MATCC	Maternal Cell Contamination, B	Yes	No	

Current Algorithm

The following are available:

-Glucose-6-Phosphate Dehydrogenase (G6PD)
Deficiency Diagnostic Algorithm

-Glucose-6-Phosphate Dehydrogenase (G6PD)
Genotyping Interpretive Algorithm

New Algorithm

For cord blood specimens that have an accompanying maternal blood specimen, maternal cell contamination studies will be performed at an additional charge.

The following are available:

-Glucose-6-Phosphate Dehydrogenase (G6PD) Deficiency Diagnostic Algorithm

-Glucose-6-Phosphate Dehydrogenase (G6PD) Genotyping Interpretive Algorithm

Current Specimen Required

Submit only 1 of the following specimens:

Specimen Type: Whole blood

Container/Tube:

Preferred: Lavender top (EDTA) or yellow top

(ACD)

Specimen Volume: 3 mL Collection Instructions:

1. Invert several times to mix blood.

2. Send whole blood specimen in original tube. Do

not aliquot.

New Specimen Required

Patient Preparation: A previous hematopoietic stem cell transplant from an allogenic donor will interfere with testing. For information about testing patients who have received a hematopoietic stem cell transplant, call 800-533-1710.

Submit only 1 of the following specimens:

Specimen Type: Whole blood

Container/Tube: Lavender top (EDTA) or yellow top (ACD)

Specimen Volume: 3 mL Collection Instructions:

1. Invert several times to mix blood.

Specimen Stability Information: Ambient (preferred) 9 days/Refrigerated 30 days

Specimen Type: Saliva

Patient Preparation: Patient should not eat, drink, smoke, or chew gum 30 minutes prior to collection.

Supplies: Saliva Swab Collection Kit (T786)

Specimen Volume: 1 Swab

Collection Instructions: Collect and send

specimen per kit instructions.

Specimen Stability Information: Ambient 30

days

2. Send whole blood specimen in original tube. **Do not aliquot**.

Specimen Stability Information: Ambient (preferred) 4 days/Refrigerated 4 days/Frozen 4 days

Additional Information:

- 1. Specimens are preferred to be received within 4 days of collection. Extraction will be attempted for specimens received after 4 days, and DNA yield will be evaluated to determine if testing may proceed.
- 2. To ensure minimum volume and concentration of DNA is met, the requested volume must be submitted. Testing may be canceled if DNA requirements are inadequate.

Specimen Type: Cord blood

Container/Tube: Lavender top (EDTA) or yellow top (ACD)

Specimen Volume: 3 mL Collection Instructions:

- 1. Invert several times to mix blood.
- 2. Send cord blood specimen in original tube. **Do not aliquot**. **Specimen Stability Information:** Ambient (preferred) 4 days/Refrigerated 4 days/Frozen 4 days

Additional Information:

- 1. Specimens are preferred to be received within 4 days of collection. Extraction will be attempted for specimens received after 4 days, and DNA yield will be evaluated to determine if testing may proceed.
- 2. To ensure minimum volume and concentration of DNA is met, the requested volume must be submitted. Testing may be canceled if DNA requirements are inadequate.
- 3. While a properly collected cord blood sample may not be at risk for maternal cell contamination, unanticipated complications may occur during collection. Therefore, maternal cell contamination studies are recommended to ensure the test results reflect that of the patient tested and are available at an additional charge. Order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Specimen Type: Saliva

Patient Preparation: Patient should not eat, drink, smoke, or

chew gum 30 minutes prior to collection. **Supplies**: Saliva Swab Collection Kit (T786)

Specimen Volume: 1 Swab

Collection Instructions: Collect and send specimen per kit

nstructions.

Specimen Stability Information: Ambient (preferred) 30

days/Refrigerated 30 days

Additional Information: Saliva specimens are acceptable but not recommended. Due to lower quantity/quality of DNA yielded from saliva, some aspects of the test may not perform as well as DNA extracted from a whole blood sample. When applicable, specific gene regions that were unable to be interrogated will be noted in the report. Alternatively, additional specimen may be required to complete testing.

Specimen Type: Extracted DNA

Container/Tube:

Preferred: Screw Cap Micro Tube, 2mL with skirted conical

base

Acceptable: Matrix tube, 1mL

	Collection
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	of 75 ng/m
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Collection Instructions:

- 1. The preferred volume is at least 100 mcL at a concentration of 75 ng/mcL.
- 2. Include concentration and volume on tube.

Specimen Stability Information: Frozen (preferred) 1 year/Ambient/Refrigerated

Additional Information: DNA must be extracted in a CLIA-certified laboratory or equivalent and must be extracted from a specimen type listed as acceptable for this test (including applicable anticoagulants). Our laboratory has experience with Chemagic, Puregene, Autopure, MagnaPure, and EZ1 extraction platforms and cannot guarantee that all extraction methods are compatible with this test. If testing fails, one repeat will be attempted, and if unsuccessful, the test will be reported as failed and a charge will be applied. If applicable, specific gene regions that were unable to be interrogated due to DNA quality will be noted in the report.

Current Days I	Performed
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Monday, Wednesday

Varies

Questions

Contact Michelle Raths, Laboratory Resource Coordinator at 800-533-1710.