

## **HLA-B\*57:01 Genotype, Pharmacogenomics, Varies**

**Test ID:** HL57R

**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 Testing Algorithm**

See [Abacavir Hypersensitivity Testing and Initial Patient Management Algorithm](#)

For additional information regarding pharmacogenomic genes and their associated drugs, see the [Pharmacogenomic Associations Tables](#)

### **New Testing Algorithm**

**Cord blood:**

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

See [Abacavir Hypersensitivity Testing and Initial Patient Management Algorithm](#)

For additional information regarding pharmacogenomic genes and their associated drugs, see the [Pharmacogenomic Associations Tables](#).

### **Current Reflex Tests**

None

### **New Reflex Tests**

| Test ID | Reporting Name                 | Available Separately | Always Performed |
|---------|--------------------------------|----------------------|------------------|
| MATCC   | Maternal Cell Contamination, B | Yes                  | No               |

### **Current Specimen Required**

Multiple genotype tests can be performed on a single specimen after a single extraction. See [Multiple Genotype Test List](#) for a list of tests that can be ordered together.

**Submit only 1 of the following specimens:**

**Specimen Type:** Whole blood  
**Container/Tube:** Lavender top (EDTA)  
**Specimen Volume:** 3 mL  
**Collection Instructions:**  
1. Invert several times to mix blood.

### **New Specimen Required**

**Patient Preparation:** A previous hematopoietic stem cell transplant or liver transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a hematopoietic stem cell or liver transplant.

**Submit only 1 of the following specimens:**

**Specimen Type:** Whole blood  
**Container/Tube:** Lavender top (EDTA)  
**Specimen Volume:** 3 mL  
**Collection Instructions:**

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

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

**Specimen Type: Saliva**

**Supplies:** Saliva Swab Collection Kit (T786)

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

**Specimen Volume:** 1 Swab

**Collection Instructions:** Collect and send specimen per kit instructions.

**Specimen Stability Information:** Ambient 30 days

**Specimen Type: Extracted DNA**

**Container/Tube:** 2 mL screw top tube

**Specimen Volume:** 100 mcL (microliters)

**Collection Instructions:**

1. The preferred volume is 100 mcL at a concentration of 50 ng/mcL.
2. Include concentration and volume on tube.

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

1. Invert several times to mix blood.

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 are 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)

**Specimen Volume:** 3 mL

**Collection Instructions:**

1. After birth, collect whole blood from the umbilical cord.
2. Invert several times to mix blood.

3. 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 are 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 instructions.

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

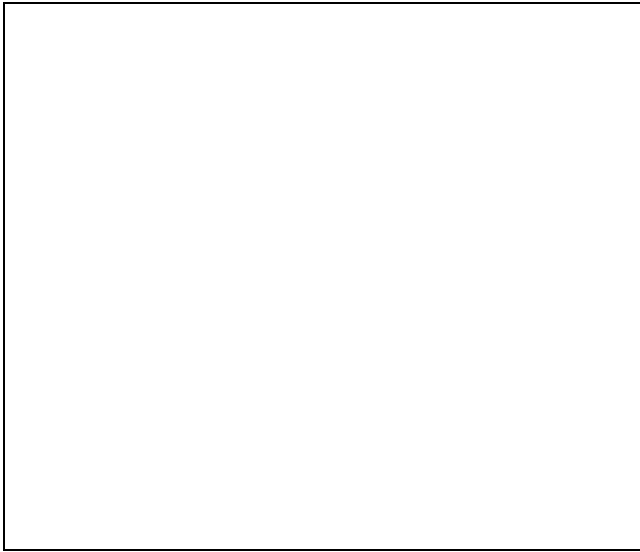
**Additional Information:** Saliva specimens are acceptable but not recommended. Due to lower quantity/quality of DNA yielded from saliva, some aspects of the test may not perform as well as DNA extracted from a whole blood sample. When applicable, specific gene regions that were unable to be interrogated 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, 2 mL with skirted conical base

**Acceptable:** Matrix tube, 1 mL



**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 Performed           |
|----------------------------------|
| Monday, Wednesday through Friday |

| New Days Performed |
|--------------------|
| Varies             |

**Questions**

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