

Carbamazepine Hypersensitivity Pharmacogenomics, Varies

Test ID: CARBR

Useful for:

Identifying individuals with increased risk of carbamazepine- or oxcarbazepine-associated cutaneous adverse reactions

Genetics Information:

Detection of the *HLA-B*15:02* allele (HLA00165) in the *HLA-B* gene (NM_005514).

Detection of the *HLA-A*31:01* allele (HLA00097) in the *HLA-A* gene (NM_001242758).

Methods:

Qualitative Allele-Specific Real-Time Polymerase Chain Reaction (PCR)

Reference Values:

An interpretive report will be provided.

Specimen Stability Information:

Specimen Type	Temperature	Time
Varies	Varies	

Specimen Requirements:

Multiple genotype tests can be performed on a single specimen after a single extraction.

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.
2. Send specimen in original tube.

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.

Additional Information: Due to lower concentration of DNA yielded from saliva, testing cannot proceed to tier 2 sequencing and will stop after tier 1 testing is complete.

Specimen Stability Information: Ambient 30 days

Specimen Type: 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 75 ng/mcL.
2. Include concentration and volume on tube.

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

Cautions:

Rare reported or unreported *HLA-A* and *HLA-B* alleles may occur and may interfere with this assay, resulting in a false-positive or false-negative call. Examples of alleles that may interfere include other *HLA-A*31* alleles (including *HLA-A *31:01:23*), *HLA-B*15:13*, *HLA-B*15:31*, *HLA-B*15:55*, *HLA-B*15:88*, *HLA-B*15:89*, *HLA-B*18:20*, *HLA-B*15:112*, *HLA-B*15:121*, *HLA-B*15:144*, and *HLA-B*15:170*. However, most of these alleles are rare and exist only in specific ethnicities, and it is not known if any of these subtypes are associated with hypersensitivity. For example, *HLA-B*15:13*, while rare, has been observed more in Asian populations than other populations.

Samples may contain donor DNA if obtained from patients who received non-leukoreduced blood transfusions or allogeneic hematopoietic stem cell transplantation (AHSCT). Results from samples obtained under these circumstances may not accurately reflect the recipient's genotype. For individuals who have received blood transfusions, the genotype usually reverts to that of the recipient within 6 weeks. The impact of AHSCT on risk of adverse cutaneous reactions is not defined in the literature.

CPT Code:

81381x2

Day(s) Setup: Monday, Wednesday through Friday

Analytic Time: 1 day

Questions

Contact your Laboratory Technologist Resource Coordinator Heather Flynn Gilmer at 800-533-1710.