NEW TEST

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HLA-B*15:02 Genotyping, Carbamazepine Hypersensitivity

3020687, HLA-B1502

Specimen Requirements:	
Patient Preparation:	
Collect:	Lavender (EDTA), pink (K2EDTA), or yellow (ACD solution A or B).
Specimen Preparation:	Transport 5 mL whole blood. (Min: 3 mL)
Transport Temperature:	Refrigerated.
Unacceptable Conditions:	Specimens collected in green (sodium or lithium heparin).
Remarks:	
Stability:	Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable
Methodology:	Polymerase Chain Reaction (PCR) / Sequence-Specific Oligonucleotide Probe Hybridization / Massively Parallel Sequencing
Note:	
CPT Codes:	81381
New York DOH Approval Status:	This test is New York DOH approved.

Interpretive Data:

Background Information for HLA-B*1502 Genotype, Carbamazepine Hypersensitivity:
Characteristics: Carbamazepine (CBZ) is an aromatic antiepileptic drug, approved for the treatment of epilepsy and trigeminal neuralgia. Rarely, CBZ can induce severe life-threatening reactions such as Stevens-Johnson syndrome (SJS) or toxic epidermal necrolysis (TEN). Symptoms usually appear within the first months of treatment, and include skin rash, hives, sores in the mouth, blistering or peeling of the skin, and erosion of the mucous membranes in the respiratory and gastrointestinal tract. The presence of HLA-B*15:02 increases risk for CBZ-induced SJS/TEN in individuals of Asian ancestry. The incidence of CBZ-induced life-threatening reactions such SJS, TEN, or hypersensitivity syndrome (HSS) is 1-10 per 10,000, which can be higher in some Asian countries.
Incidence: HLA-B*15:02 allele frequency varies by ethnicity, with highest incidence in Asians: 10.2 percent in Han Chinese, 10 percent in Taiwanese (18 percent in indigenous Puyuma), greater than 5 percent in the populations of Hong Kong, Thailand, Malaysia, Vietnam, Philippines, India (Khandesh and West Bhil), and Indonesia. Frequency is low in African Americans (0.1-1 percent) and less than 0.1 percent in Caucasians.
Cause: In patient of Asian descent, CBZ-induced SJS/TEN is strongly associated with the presence of HLA-B*15:02 allele. The mechanism is immune mediated and involves drug-induced changes in peptide presentation by HLA-B*15:02, which allows for the activation of self-reactive T lymphocytes. Activated immune cells contribute to the cellular death of keratinocytes in the skin, which causes the epidermal destruction and detachment of the skin seen in SJS/TEN.
Alleles tested: HLA-B*15:02 allele. Other members of the HLA B75 serogroup detected by this

assay can also be associated with carbamazepine-induced SJS/TEN.
Clinical Sensitivity and Specificity: 80-97 percent and 99 percent, respectively in populations where the HLA-B*15:02 allele is common.
Methodology: PCR followed by sequence-specific oligonucleotide probe hybridization of HLA-B locus.
Analytical Sensitivity and Specificity: Greater than 99 percent.
Limitations: Other genetic and non-genetic factors that influence carbamazepine hypersensitivity are not evaluated. Other rare, or novel alleles may occur which may lead to false positive or false negative results.

Test systems were developed and their performance characteristics determined by the H&I laboratory at the University of Utah Health, under the accreditation guidelines from the American Society for Histocompatibility and Immunogenetics (ASHI).

Reference Interval:

Refer to report

HOTLINE NOTE: Refer to the Hotline Test Mix for interface build information.