NEW TEST

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HLA-B*58:01 Genotyping, Allopurinol Hypersensitivity

3020683, HLA-B5801

Specimen Requirements:	
Patient Preparation:	
Collect:	Lavendar (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:

Characteristics: Allopurinol is the most commonly used drug for the treatment of hyperuricemia and gout. It inhibits xanthine oxidase, a key enzyme involved in uric acid formation. However, allopurinol is one of the most common causes of life-threatening severe cutaneous adverse reactions (SCAR), which include drug hypersensitivity syndrome, Stevens-Johnson syndrome (SJS), and toxic epidermal necrolysis (TEN). The presence of HLA-B*58:01 allele shows strong association with allopurinol-induced SCAR, including TEN and SJS. Although allopurinol-induced SCAR is rare, with an estimated risk of 0.1-0.4 percent in allopurinol users, the severity can be high, with a mortality rate of up to 25 percent. Symptoms include rash, combined with eosinophilia, leukocytosis, fever, hepatitis, and progressive kidney failure. Due to the severity of adverse reactions, it is recommended to test for the HLA-B*58:01 allele prior to initiation of the drug.
Incidence: HLA-B*58:01 allele frequency varies by ethnicity. In the U.S. population, the highest incidence at 5.3 percent is found in Asians, 3.8 percent in African Americans, 1.45 percent in Native Hawaiians or Pacific Islanders, 1.35 percent in Hispanics, 1.19 percent in American Indians or Alaska Natives, and 0.8 percent in Caucasians. Frequencies may be higher in other countries, up to 20 percent in Singapore, Taiwan, and among Han Chinese, 15.4 percent in India, 14.2 percent in Hong Kong, 12 percent in China and Korea, 11 percent in Indonesia.
Cause: Allopurinol-induced SCAR, including SJS and TEN, is strongly associated with the presence of one or two copies of HLA-B*58:01 allele. The mechanism is immune mediated and involves direct interactions between the allopurine metabolite oxypurinol, and HLA-B*58:01, which may result in drug-induced changes in peptide presentation, allowing activation of self-reactive T lymphocytes.

Alleles tested: HLA-B*58:01 allele.
Clinical Sensitivity and Specificity: 71 percent sensitivity and 92 percent specificity, overall mean values from pooled populations (Yu KH et al, Int J Rheum Dis 2017). Higher in populations with increased HLA-B*58:01 allele frequency.
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 allopurinol 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.