NEW TEST

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Narcolepsy HLA-DQ Genotyping (HLA-DQB1*06:02)

3017170, HLANARCO

Specimen Requirements:	
Patient Preparation:	
Collect:	Lavender (EDTA). Also acceptable: Yellow (ACD solution A).
Specimen Preparation:	Transport 3 mL whole blood. (Min: 1 mL)
Transport Temperature:	Refrigerated.
Unacceptable Conditions:	Specimens collected in yellow (ACD solution B). Clotted, grossly hemolyzed, or heparinized specimens.
Remarks:	
Stability:	Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable.
Methodology:	Massively Parallel Sequencing Sequence-Specific Oligonucleotide Probe Hybridization Polymerase Chain Reaction (PCR)
Performed:	Mon-Fri
Reported:	8-15 days
Note:	
CPT Codes:	81382
New York DOH Approval Status:	This test is New York DOH approved.

Interpretive Data:

Background information for Narcolepsy (*HLA-DQB1**06:02) Genotyping:

Characteristics: Narcolepsy is a chronic neurological sleep disorder that manifests in excessive daytime sleepiness and difficulty in maintaining wakefulness. Narcolepsy type 1 is associated with cataplexy (the sudden loss of muscle tone triggered by strong emotions). Additionally, disturbed nighttime sleep, sleep paralysis, and hypnagogic hallucinations (occurring in the period between sleep and wakefulness) are common.

Incidence: Varies, depending on ethnicity. It affects 0.02-0.05% of the populations in the US and Europe, it is most common in Japan (0.16-0.18%).

Inheritance: Multifactorial.

Cause: The *HLA-DQB1**06:02 allele is strongly associated with narcolepsy, but by itself is not causative. Homozygosity for DQB1*06:02 allele doubles the risk, compared to heterozygous individuals.

Alleles Tested: *HLA-DQB1* alleles.

Clinical Sensitivity: 85-95 percent depending on ethnicity. Greater than 98% of affected Caucasians with cataplexy have the *HLA-DQB1**06:02 allele.

Clinical Specificity: Less than 1 percent; 15-25 percent of unaffected Caucasians carry the *HLA-DQB1**06:02 allele.

Methodology: Polymerase Chain Reaction/Massively Parallel Sequencing, or Polymerase Chain Reaction/Sequence-Specific Oligonucleotide Probe Hybridization

Analytical Sensitivity and Specificity: 99 percent.

Limitations: Rare diagnostic errors may occur due to primer site mutations. Other genetic and nongenetic factors that influence narcolepsy disease are not evaluated. In cases where an HLA allele cannot be resolved unambiguously, the allele assignment will be reported as the most common, based on allele frequencies from the common, intermediate, and well-documented alleles catalogue version 3.0.0 (Hurley CK et al, 2020).

This test was developed and its performance characteristics determined by the Histocompatibility & Immunogenetics laboratory at the University of Utah Health. It has not been cleared or approved by the US Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes. It should not be regarded as investigational or for research. Histocompatibility & Immunogenetics laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA-88) as qualified to perform high complexity clinical laboratory testing.

Performed at: Histocompatibility & Immunogenetics Laboratory, University of Utah Health, 417 Wakara Way, Suite 3220, Salt Lake City, UT 84108.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

Reference Interval:

By report

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