Narcolepsy (HLA-DQB1*06:02) Genotyping
ARUP test code 2005023

Narcolepsy - Specimen
Whole Blood

Narcolepsy (HLA-DQB1*06:02) Positive *

Result: Positive for HLA-DQB1*06:02

INTERPRETIVE INFORMATION: The HLA-DQB1*06:02 allele, which is strongly associated with narcolepsy, was detected. This result is supportive of a clinical diagnosis of narcolepsy, but by itself does not establish a diagnosis. Medical screening and management of this individual should rely on clinical findings.

This result has been reviewed and approved by Hunter Best, Ph.D.
BACKGROUND INFORMATION: Narcolepsy (HLA-DQB1*06:02) Genotyping

CHARACTERISTICS: Narcolepsy is a sleep disorder associated with invalidating excessive daytime sleepiness and 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: Narcolepsy affects approximately 1 in 2,000 individuals.

INHERITANCE: Multifactorial.

CAUSE: The HLA-DQB1*06:02 allele is strongly associated with narcolepsy, but by itself is not causative. Recent studies indicate HLA-DRB1*15 is not associated with narcolepsy.

MUTATIONS TESTED: HLA-DQB1*06:02 allele.

CLINICAL SENSITIVITY: 85-95 percent depending on ethnicity. Greater than 99 percent of affected Caucasians with cataplexy have the HLA-DQB1*06:02 allele.

CLINICAL SPECIFICITY: Less than 1 percent; 15-25 percent of unaffected Caucasians have the HLA-DQB1*06:02 allele.

METHODOLOGY: PCR with melting curve analysis.

ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.

LIMITATIONS: This methodology will not differentiate between individuals who are heterozygous (one copy) and those who are homozygous (two copies) for the HLA-DQB1*06:02 allele. Rare diagnostic errors may occur due to primer site mutations. Alleles other than HLA-DQB1*06:02 are not identified. Other genetic and non-genetic factors that influence narcolepsy are not evaluated.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online at www.aruplab.com.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

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