

500 Chipeta Way, Salt Lake City, Utah 84108-1221

phone: 801-583-2787, toll free: 800-522-2787

Tracy I. George, MD, Chief Medical Officer

Patient Age/Sex: 39 years Male

Specimen Collected: 08-Mar-22 10:46

Narcolepsy (HLA-DQB1 06:02) | Received: 08-Mar-22 10:46 Report/Verified: 10-Mar-22 13:58

Genotyping

Procedure	Result	Units	Reference Interval
Narcolepsy -Specimen	Whole Blood		
Narcolepsy (HLA-DQB1*06:02)	Negative ^{f1 i1}		

Result Footnote

f1: Narcolepsy (HLA-DQB1*06:02)

Result: Negative for HLA-DQB1*06:02

INTERPRETIVE INFORMATION: The HLA-DQB1*06:02 allele, which is strongly associated with narcolepsy, was not detected. For this patient, the likelihood of a diagnosis of narcolepsy is reduced but may not be eliminated; approximately 10 percent of affected patients may not have the HLA-DQB1*06:02 allele. Medical screening and management of this individual should rely on clinical findings.

This result has been reviewed and approved by Eszter Lazar-Molnar, Ph.D.

Test Information

i1: Narcolepsy (HLA-DQB1*06:02)

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: Polymerase chain reaction (PCR) and fluorescence monitoring.

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.

*=Abnormal, #=Corrected, C=Critical, f=Result Footnote, H-High, i-Test Information, L-Low, t-Interpretive Text, @=Performing lab

Unless otherwise indicated, testing performed at:

ARUP Laboratories

500 Chipeta Way, Salt Lake City, UT 84108

Laboratory Director: Tracy I. George, MD

ARUP Accession: 22-067-900091

Report Request ID: 15080557

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Test Information

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This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

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

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