Client: ARUP Example Report Only
500 Chipeta Way
Salt Lake City, UT 84108
UNITED STATES

Physician: TEST, TEST

Patient: Patient, Example

DOB	1/10/1975
Sex:	Female
Patient Identifiers:	55605
Visit Number (FIN):	55993
Collection Date:	12/20/2023 10:01

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

ARUP test code 3017170

HLA-DQB1, Allele 1	02:01
HLA-DQB1, Allele 2	02:01
Narcolepsy HLA Interpretation	See Note

Negative for HLA-DQB1*06:02
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 completely eliminated. Up to 10 percent of affected narcolepsy type 1 patients, and 50-80 percent of narcolepsy type 2 patients may not carry the HLA-DQB1*06:02 allele. Medical screening and management of this individual should rely on clinical findings.

BACKGROUND INFORMATION: Narcolepsy Genotyping (HLA-DQB1*06:02)
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

H=High, L=Low, *=Abnormal, C=Critical

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.
CLIA Number: 46D0679773

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
HLA-DQB1, Allele 1	23-354-103967	12/20/2023 10:01:00 AM	12/20/2023 10:52:27 AM	12/20/2023 10:58:00 AM
HLA-DQB1, Allele 2	23-354-103967	12/20/2023 10:01:00 AM	12/20/2023 10:52:27 AM	12/20/2023 10:58:00 AM
Narcolepsy HLA Interpretation	23-354-103967	12/20/2023 10:01:00 AM	12/20/2023 10:52:27 AM	12/20/2023 10:58:00 AM

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