

Overview

Useful For

Ruling out a diagnosis of narcolepsy

Method Name

Polymerase Chain Reaction (PCR)/Sequence-Specific Oligonucleotide Probes (SSO)

NY State Available

Yes

Specimen

Specimen Type

Whole Blood ACD-B

Specimen Required

Container/Tube:

Preferred: Yellow top (ACD solution B)

Acceptable: Yellow top (ACD solution A), lavender top (EDTA)

Specimen Volume: 6 mL

Collection Instructions: Send whole blood specimen in original vial. **Do not aliquot.**

Additional Information: Specimen acceptability is based on extracted DNA concentration and not sample age.

Specimen Minimum Volume

3 mL

Reject Due To

Extracted DNA	Reject
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Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole Blood ACD-B	Refrigerated (preferred)		
	Ambient		

Clinical & Interpretive

Clinical Information

Narcolepsy is a neurological condition affecting about 0.02% of African American, White, and Japanese individuals. It is characterized by excessive daytime somnolence and abnormal rapid eye movement (REM) sleep. Cataplexy (weakness precipitated by emotions, especially laughter) is present in 64% to 79% of patients with narcolepsy.

Studies have identified *DQB1*06:02* as a useful marker of narcolepsy. *DQB1*06:02* is found in 90% to 95% of African American, White, and Japanese patients with narcolepsy who also have cataplexy (narcolepsy type 1) but only in 45% to 50% of patients with narcolepsy without cataplexy (narcolepsy type 2). It must also be clearly understood that about 25% of normal people have this gene.

Because *DQB1*06:02* is present in the normal population, no test for an *HLA* gene constitutes a test for narcolepsy. A more reliable approach would be to consider that, in an appropriate patient who has cataplexy, the absence of the strongly associated *DQB1*06:02* provides good evidence that the patient does **not** have narcolepsy. However, its absence does not rule-out narcolepsy without cataplexy (narcolepsy type 2).

Reference Values

An interpretive report will be provided.

Interpretation

If *DQB1*06:02* is not detected, the narcolepsy-associated antigen test result will be reported as negative for *DQB1*06:02*.

If the allele is detected, the result will be reported as positive for *DQB1*06:02*.

Cautions

Based on the catalog of common, intermediate, and well-documented alleles in the world population,(1) certain intermediate or common alleles in some ethnicities may not be resolved.

Clinical Reference

1. Hurley CK, Kempenich J, Wadsworth K, et al. Common, intermediate and well-documented HLA alleles in world populations: CIWD version 3.0.0. *HLA*. 2020;95(6):516-531. doi:10.1111/tan.13811
2. Mignot E, Lin X, Arrigoni J, et al. *DQB1*0602* and *DQA1*0102* (DQ1) are better markers than DR2 for narcolepsy in Caucasian and Black Americans. *Sleep*. 1994;17(8 Suppl):S60-S67
3. Chabas D, Taheri S, Renier C, Mignot E. The genetics of narcolepsy. *Annu Rev Genomics Hum Genet*. 2003;4:459-483
4. Andlauer O, Moore H 4th, Hong SC, et al. Predictors of hypocretin (orexin) deficiency in narcolepsy without cataplexy. *Sleep*. 2012;35(9):1247-1255F
5. Bassetti CLA, Adamantidis A, Burdakov D, et al. Narcolepsy - clinical spectrum, aetiopathophysiology, diagnosis and treatment. *Nat Rev Neurol*. 2019;15(9):519-539. doi:10.1038/s41582-019-0226-9
6. Capittini C, De Silvestri A, Terzaghi M, et al. Correlation between HLA-DQB1*06:02 and narcolepsy with and without cataplexy: approving a safe and sensitive genetic test in four major ethnic groups. A systematic meta-analysis. *Sleep Med*. 2018;52:150-157
7. Miyagawa T, Tokunaga K. Genetics of narcolepsy. *Hum Genome Var*. 2019;6:4. doi:10.1038/s41439-018-0033-7

Performance

Method Description

This assay applies Luminex technology to the reverse sequence specific oligonucleotide DNA typing method. First, target DNA is polymerase chain reaction (PCR)-amplified using a group-specific primer. The PCR product is biotinylated, which allows it to be detected using r-phycoerythrin-conjugated streptavidin. The PCR product is denatured and allowed to rehybridize to complementary DNA probes conjugated to fluorescently coded microspheres. A flow analyzer identifies the fluorescent intensity of phycoerythrin on each microsphere. The *HLA* class II allele or allele groups of the sample are determined by the positive and negative bead identified using a computer software program. The assignment of the human leukocyte antigen (HLA) typing is based on the reaction pattern compared to patterns associated with published *HLA* gene sequences.(Package insert: LABType SSO Typing Tests. One Lambda; Version 04, 11/11/2019)

PDF Report

No

Day(s) Performed

Monday through Friday

Report Available

3 to 8 days

Specimen Retention Time

14 days

Performing Laboratory Location

Mayo Clinic Laboratories - Rochester Main Campus

Fees & Codes

Fees

- Authorized users can sign in to [Test Prices](#) for detailed fee information.
- Clients without access to Test Prices can contact [Customer Service](#) 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact [Customer Service](#).

Test Classification

This test has been modified from the manufacturer's instructions. Its performance characteristics were determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

81376-HLA Class II typing, low resolution (eg, antigen equivalents); one locus (eg, HLA-DRB1/3/4/5, -DQB1, -DQA1, -DPB1, or -DPA1), each

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
NARC	Narcolepsy Associated Ag, B	63558-1

Result ID	Test Result Name	Result LOINC® Value
NARC_	Narcolepsy Associated Ag Result	63558-1
NARCC	Interpretation	50595-8