Overview

Useful For

Assessing risk of celiac disease

Testing Algorithm

The following algorithms are available in Special Instructions:

-Celiac Disease Comprehensive Cascade

-Celiac Disease Diagnostic Testing Algorithm

-Celiac Disease Gluten-Free Cascade

-Celiac Disease Routine Treatment Monitoring Algorithm

-Celiac Disease Serology Cascade

Special Instructions

- Celiac Disease Diagnostic Testing Algorithm
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Method Name

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

NY State Available

Yes

Specimen

Specimen Type

Whole Blood ACD-B

Advisory Information

Cascade testing is recommended for celiac disease. Cascade testing ensures that testing proceeds in an algorithmic fashion. The following cascades are available; select the appropriate one for your specific patient situation.

-CDCOM / Celiac Disease Comprehensive Cascade: complete testing including HLA DQ

-CDSP / Celiac Disease Serology Cascade: complete testing excluding HLA DQ

-CDGF / Celiac Disease Gluten-Free Cascade: for patients already adhering to a gluten-free diet

To order individual tests, see Celiac Disease Diagnostic Testing Algorithm in Special Instructions.

Specimen Required

Container/Tube: Yellow top (ACD solution B)

Specimen Volume: 6 mL

Collection Instructions: Do not transfer blood to other containers.

Forms

If not ordering electronically, complete, print, and send a Gastroenterology and Hepatology Client Test Request (T728) with the specimen.

Specimen Minimum Volume

3 mL

Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

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

Clinical and Interpretive

Clinical Information

Celiac disease (gluten-sensitive enteropathy) is mediated by T lymphocytes in patients with genetic susceptibility. This genetic association is with certain HLA genes in the class II region (DQ alpha 1, DQ beta 1).

Reference Values

An interpretive report will be provided.

Interpretation

Most (90%-95%) patients with celiac disease have 1 or 2 copies of HLA-DQ2 haplotype (see below), while the remainder have HLA-DQ8 haplotype. Rare exceptions to these associations have been occasionally seen. In 1 study of celiac disease, only 0.7% of patients with celiac disease lacked the HLA alleles mentioned above. Results are reported as permissive, nonpermissive, or equivocal gene pairs.

It is important to realize that these genes are also present in about 20% of people without celiac disease. Therefore, the mere presence of these genes does not prove the presence of celiac disease or that genetic susceptibility to celiac disease is present.

The HLA-DQ molecule is composed of two chains: DQ alpha (encoded by *HLA-DQA1* gene) and DQ beta (encoded by *HLA-DQB1* gene). HLA-DQ typing can be performed by serological or molecular methods. Currently most laboratories perform typing by molecular methods. HLA-DQ2 and DQ8 as typed by serology are usually based on the molecular typing of the DQB1 chain only. The current molecular method allows typing for both the DQB1 and DQA1 chains and this has shown that there are different haplotypes of HLA-DQ2 and DQ8. Typing of these haplotypes is important in celiac disease as they carry different risk association.

There are 2 common haplotypes of DQ2:

1. *DQA1*05:01* with *DQB1*02:01* also called DQ2.5 in celiac literature

2. *DQA1*02:01* with *DQB1*02:02* also called DQ2.2 in celiac literature

A single haplotype (heterozygote) of DQ2.5 is permissive for presence of celiac genes. However, only a double haplotype (homozygous) of DQ2.2 is permissive for presence of celiac genes. There are few reports where a single haplotype of DQ2.2 is considered to be an equivocal risk. In some cases the DQ2.2 haplotype maybe present with a DQ7.5 haplotype (*DQA1*05:05* with *DQB1*03:01*). In this case a DQ2.5 molecule can be formed by the combination of *DQB1*02:02* from 1 chromosome and *DQA1*05:05* from the other chromosome. These cases fall in the same category as the DQ2.5 heterozygote.

There are 3 common haplotypes of DQ8:

1. *DQA1*03:01* with *DQB1*03:02*

2. *DQA1*03:02* with *DQB1*03:02*

3. *DQA1*03:03* with *DQB1*03:02*

Any single haplotype (heterozygote) of DQ8 is permissive for celiac.

Therefore, the gene pairs permissive for celiac are:

1. Heterozygote (single copy)

-*DQA1*05:XX* with *DQB1*02:01*

-*DQA1*05:XX* with *DQB1*02:02*

-*DQA1*03:XX* with *DQB1*03:02*

2. Homozygous (2 copies)

-*DQA1*02:01* with *DQB1*02:02*

Gene pairs equivocal for celiac are

1. Heterozygote (single copy)

-*DQA1*02:01* with *DQB1*02:02*

2. Rare alleleÃ¢â‚¬â„¢s types of DQ2 and DQ8 other than those listed above

All other gene pair combinations are considered non-permissive for celiac.

Cautions

No significant cautionary statements

Clinical Reference

Polvi A, Arranz E, Fernandez-Arequero M, et al: HLA-DQ2-negative celiac disease in Finland and Spain. Hum

Immunol 1998 Mar;59(3):169-175

Performance

Method Description

LABType applies Luminex technology to the reverse sequence-specific oligonucleotide (SSO) DNA typing method. First, target DNA is 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 is determined by the positive and negative bead ID's using a computer software program. The assignment of the HLA typing is based on the reaction pattern compared to patterns associated with published HLA gene sequences.(Package insert: One Lambda, LABType SSO Typing)

PDF Report

No

Day(s) and Time(s) Test Performed

Monday through Friday; 7:30 a.m.-5 p.m.

Analytic Time

5 days

Maximum Laboratory Time

9 days

Specimen Retention Time

Whole Blood 7 days; Extracted DNA 2 months

Performing Laboratory Location

Rochester

Fees and 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 Regional Manager. For assistance, contact Customer Service.

Test Classification

This test has been cleared or approved by the U.S. Food and Drug Administration and is used per manufacturer's instructions. Performance characteristics were verified by Mayo Clinic in a manner consistent with CLIA requirements.

CPT Code Information

81376 x 2-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 Definition: CELI

Celiac Associated HLA-DQ Typing

Test ID	Test Order Name	Order LOINC Value
CELI	Celiac Associated HLA-DQ Typing	94492-6

Result ID	Test Result Name	Result LOINC Value
DQA	DQ alpha 1	44728-4
DQB	DQ beta 1	43291-4
CELIG	Celiac gene pairs present?	48767-8
CELIC	Interpretation	69048-7