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
- Celiac Disease Comprehensive Cascade
- Celiac Disease Gluten-Free Cascade
- Celiac Disease Routine Treatment Monitoring Algorithm
- Celiac Disease Serology Cascade

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
**Test Definition: CELI**
Celiac Associated HLA-DQ Typing

**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)](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**

<table>
<thead>
<tr>
<th>Specimen Type</th>
<th>Temperature</th>
<th>Time</th>
<th>Special Container</th>
</tr>
</thead>
<tbody>
<tr>
<td>Whole Blood ACD-B</td>
<td>Refrigerated (preferred)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Ambient</td>
<td></td>
</tr>
</tbody>
</table>

**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 allele 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**

Test Definition: CELI
Celiac Associated HLA-DQ Typing


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, approved or is exempt 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

<table>
<thead>
<tr>
<th>Test ID</th>
<th>Test Order Name</th>
<th>Order LOINC Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>CELI</td>
<td>Celiac Associated HLA-DQ Typing</td>
<td>94492-6</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Result ID</th>
<th>Test Result Name</th>
<th>Result LOINC Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>DQA</td>
<td>DQ alpha 1</td>
<td>94495-9</td>
</tr>
<tr>
<td>DQB</td>
<td>DQ beta 1</td>
<td>53938-7</td>
</tr>
<tr>
<td>CELIG</td>
<td>Celiac gene pairs present?</td>
<td>48767-8</td>
</tr>
<tr>
<td>CELIC</td>
<td>Interpretation</td>
<td>69048-7</td>
</tr>
</tbody>
</table>