Celiac Disease Testing

Introduction

Celiac disease testing is addressed by this guideline.

Procedures addressed

The inclusion of any procedure code in this table does not imply that the code is under management or requires prior authorization. Refer to the specific Health Plan’s procedure code list for management requirements.

<table>
<thead>
<tr>
<th>Procedures addressed by this guideline</th>
<th>Procedure codes</th>
</tr>
</thead>
<tbody>
<tr>
<td>HLA Class II Typing- low resolution, multiple alleles</td>
<td>81376</td>
</tr>
<tr>
<td>HLA Class II Typing- low resolution, one allele or allele group</td>
<td>81377</td>
</tr>
<tr>
<td>HLA Class II Typing- high resolution, multiple alleles</td>
<td>81382</td>
</tr>
<tr>
<td>HLA Class II Typing- high resolution, one allele or allele group</td>
<td>81383</td>
</tr>
</tbody>
</table>

What is Celiac disease

Definition

Celiac disease is an immune-mediated disorder that mainly affects the digestive tract.1-4

Symptoms

Symptoms include diarrhea, constipation, vomiting, abdominal pain and bloating, growth problems, iron deficiency anemia, osteoporosis, and other complications of malabsorption.1-4

Prevalence

Celiac disease affects infants, children, and adults and can present at any age. It affects about 1 in every 100 people in the U.S.2,3
**Cause**

Celiac disease is caused by multiple factors, including environmental trigger in a genetically predisposed person, upon exposure to dietary gluten, which is a protein molecule found in wheat, barley and rye.¹⁻⁴

**Diagnosis**

An initial diagnosis of celiac disease is highly suspected based on serologic testing and is confirmed by finding characteristic changes on intestinal biopsy. Intestinal biopsy remains the gold standard for making a diagnosis of celiac disease.¹⁻⁴

**Increased risk**

Patients with certain medical conditions and relatives of people with celiac disease are known to have an increased risk of developing the condition.²⁻³

**Test information**

**Introduction**

Two HLA haplotypes are associated with celiac disease: HLA-DQ2 and HLA-DQ8, and are determined by molecular testing of the genes HLA-DQA1 and HLA-DQB1. These haplotypes are present in about 30-40% of the general population, but more than 99% of patients with celiac disease have one or more of these variants¹. If a person suspected of having celiac disease is found not to have one of these markers, the diagnosis can be essentially excluded.²⁻⁴

**Guidelines and evidence**

**Introduction**

This section includes relevant guidelines and evidence pertaining to Celiac disease testing.

**Guidelines**

Consensus-based guidelines from the American Gastroenterological Association (2006), the National Institutes of Health (2005), North American Society for Pediatric Gastroenterology, Hepatology and Nutrition (2005) and the 2013 American College of Gastroenterology Practice Guidelines state that HLA typing for celiac disease should be used as outlined in this table.²⁻⁵
<table>
<thead>
<tr>
<th>Test type</th>
<th>Use</th>
<th>Level of recommendation</th>
</tr>
</thead>
<tbody>
<tr>
<td>Human leukocyte antigen DQ2/DQ8 testing</td>
<td>Do not use routinely in the initial diagnosis of Celiac disease</td>
<td>Strong recommendation, moderate level of evidence</td>
</tr>
<tr>
<td>Human leukocyte antigen DQ2/DQ8 genotyping</td>
<td>Use to effectively rule out the disease in selected clinical situations</td>
<td></td>
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</tbody>
</table>

The European Society for Pediatric Gastroenterology, Hepatology, and Nutrition Guidelines for the Diagnosis of Coeliac Disease (2012) suggest screening in asymptomatic children and adolescents who are at risk for the disorder. Included in this group are individuals with type I diabetes, Down Syndrome, autoimmune thyroid disease, Turner Syndrome, Williams Syndrome, selective IgA deficiency, autoimmune liver disease, and first degree relatives diagnosed with celiac disease.

The World Gastroenterology Organisation Global Guidelines on Celiac disease (2016) state that for first degree relatives of individuals with celiac disease, no further work-up is necessary in those who test negative for HLA-DQ2/8.

**Criteria**

**Introduction**

Requests for Celiac disease testing are reviewed using these criteria.

**Criteria**

Testing may be considered in individuals who meet the following criterion:

- Celiac disease is in the differential diagnosis, but the individual has had ambiguous or indeterminate results from serology and biopsy.

**References**

**Introduction**

These references are cited in this guideline.


