COELIAC DISEASE TEST REPORT

Client Information

Donor Name: John Doe

Sample Type: A self-collected buccal swab sample was received from the individual named above; the sample was received in good condition, labelled with a unique barcode and was deemed suitable for the test. All data above has been provided by the customer.

Explanation of the test

A buccal swab sample was successfully self-collected from the individual named above and placed into a swab envelope which was returned to the laboratory for the purpose of conducting a lab-based real-time PCR (RT-PCR) genetic test for predisposition to coeliac disease. The human leukocyte antigen (HLA)-typing assay detects the presence or absence of alleles associated with coeliac disease within the sample.

Results of the test

Based on DNA analysis, the table below shows the risk-factor and haplotype associated with the sample:

<table>
<thead>
<tr>
<th>Risk</th>
<th>Haplotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>High</td>
<td>DQ2.5</td>
</tr>
</tbody>
</table>

The table below shows the specific HLA alleles tested for and whether they were present or absent in the sample:

<table>
<thead>
<tr>
<th>HLA-DQA1*05</th>
<th>HLA-DQB1*02</th>
<th>HLA-DQB1*03 02</th>
<th>HLA-DQA1*02</th>
<th>HLA-DQA1*03</th>
<th>No HLA-DQB1*02</th>
</tr>
</thead>
<tbody>
<tr>
<td>Present</td>
<td>Present</td>
<td>Absent</td>
<td>Absent</td>
<td>Present</td>
<td>Absent</td>
</tr>
</tbody>
</table>

The combination of the HLA alleles that are present or absent determines the haplotype that an individual has; the haplotype is linked to the risk of having or developing coeliac disease.
Understanding your result

The coeliac disease DNA test determines the presence or absence of HLA variants which are known to be associated with coeliac disease susceptibility, including the DQ2.5 alleles (HLA-DQA1*05, HLA-DQB1*02) and the DQ8 allele (HLA-DQB1*0302). These variants are thought to increase the risk of coeliac disease by causing receptors in the immune system to preferentially bind to a gluten component and initiating an autoimmune response.

The presence of coeliac disease-associated HLA alleles implies an increased risk for coeliac disease, however it is not diagnostic of the illness, as only a subset of individuals with these alleles will develop coeliac disease. An individual who is a first-degree relative (parent, sibling or child) of someone with coeliac disease has a greater risk of also developing coeliac disease.

Individuals who do not possess the coeliac disease-associated alleles (HLA-DQA1*05, HLA-DQB1*02 and HLA-DQB1*0302) have almost no lifetime risk of developing coeliac disease, regardless of whether they ingest gluten.

Recommendations

If you are experiencing symptoms of coeliac disease, and have received a risk result (low, moderate, or high), you should consult your doctor about conducting further tests to confirm or rule out a diagnosis of coeliac disease. A doctor may also suggest eliminating gluten from your diet, to see if your symptoms improve.

Limitations of the test

This test cannot be used to diagnose an individual with coeliac disease. You may, however, wish to share this information with your doctor, particularly if you are experiencing symptoms of coeliac disease. Your doctor will be able to recommend your next steps and provide dietary advice if they think this is appropriate.