Report Date: 27 March 2024
Barcode Number: ABC1234
Reporting Scientist: A.N. Other

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CELIAC DISEASE TEST REPORT

Client Information

Donor Name: John Doe
Date of Birth: 01 February 1995 **Sample Collection Date:** 27 March 2024

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 celiac disease. The human leukocyte antigen (HLA)-typing assay detects the presence or absence of alleles associated with celiac 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:

Risk	Haplotype
High	DQ2.5

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

High Risk Alleles			Low Risk Alleles		
HLA-DQA1*05	HLA-DQB1*02	HLA-DQB1*03:02	HLA-DQA1*02	HLA-DQA1*03	Other HLA-DQB1
Present	Present	Absent	Absent	Present	Absent

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 celiac disease.

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Understanding your result

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

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

Individuals who do not possess the high risk celiac disease-associated alleles (where alleles are not present at HLA-DQA1*05, HLA-DQB1*02 and/or HLA-DQB1*0302) have almost no lifetime risk of developing celiac disease, regardless of whether they ingest gluten.

Recommendations

If you are experiencing symptoms of celiac 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 celiac disease. A doctor may also suggest eliminating gluten from your diet, to see if your symptoms

Limitations of the test

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

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