

Celiac Disease GenotypR™ INFORMED CONSENT

1. What is celiac disease? Celiac disease (CD) is a digestive disease that damages the small intestine and interferes with absorption of nutrients from food. People who have CD cannot tolerate a protein called gluten, found in wheat, rye, and barley. When people with CD eat foods or use products containing gluten, their immune system responds by damaging the small intestine. Because the body's own immune system causes the damage, CD is considered an autoimmune disorder. Recent findings estimate about 2 million people in the United States have CD, or about 1 in 133 people.

2. What causes Celiac Disease? The cause of CD is unknown. Current research indicates that it is strongly associated with a group of genes on Chromosome 6. These genes (HLA DQ2 and HLA DQ8) are involved in the regulation of the body's immune response to the gluten. About 40% of the general population has the DQ2 and/or DQ8 markers, whereas more than 95% of patients with CD have one or both genetic markers. There is clear evidence of a family tendency toward it. 5-10% of the first-degree relatives (parents, children, and siblings) of diagnosed celiac patients may develop CD. The disease affects both sexes, and it can begin at any age, from infancy (as soon as cereal grains are introduced) to later life (even though the individual has consumed cereal grains all along).

3. What is the purpose of this test and what are its limitations? This test detects the presence of two genes, HLA DQ2 and HLA DQ8. The diagnosis of CD is consistent with the presence of HLA DQ2 and/or DQ8 in patients with CD. When both HLA markers are negative, it is very unlikely that CD is present. If clinical symptoms are not present in patients with HLA DQ2 and/or DQ8, additional analysis of immunoassays or intestinal biopsy is recommended to support a diagnosis of CD. HLA testing is not a screening test for CD; rather the value of the test lies in the ability to exclude disease when a negative result occurs. In individuals with symptoms who have not had a biopsy to diagnose celiac disease, but have been on the gluten-free diet for a significant period of time, the gene test is often the only way to determine if symptoms could possibly be related to celiac disease. In this situation, a negative gene test would in most cases indicate that symptoms are not the result of celiac disease. A positive gene test, however, does not diagnose the disease but increases the likelihood that it is present.

4. What is required to perform this test? You or your child will be asked to provide 5 mL of blood, which is equal to about one tablespoon. DNA will be extracted from this blood sample and tested. The only discomfort is the stick of the needle in the arm. A small bruise at the site of the needle puncture might be experienced. You will also be asked to provide information regarding your or your child's medical history, which is necessary for proper interpretation of the test result. In the unlikely event that you or your child should be injured in the course of being tested, your physician will provide any necessary medical care. However, you would be expected to bear the cost of such medical care. Compensation will not be provided in the event of any injury.

5. Is there a cost for this test? This is a routine clinical laboratory test and the results may aid in your or your child's diagnosis; thus, you or your health insurer will be billed for this procedure.

6. What will happen to the DNA once the test is complete? The original blood sample will be discarded at the end of the testing process or stored not more than 60 days. The DNA will be retained for a minimum of 6 months. In some circumstances, a patient's DNA may be used anonymously as a negative or positive control sample in future testing, but, in this circumstance, all identifiers will be removed prior to re-testing and the DNA sample and results obtained will remain anonymous.

I understand and agree that my DNA remaining after testing may be stored for up to 6 months should additional testing be required. *Please initial.*

7. How will I obtain results from this test? DNA testing and interpretation of results are complex. The information from this test will be provided in the form of a written report to your physician who will inform you of the results. The laboratory will not provide results directly to patients. Your physician may suggest genetic counseling prior to performing this test or if your results are abnormal. To the extent permitted by law, all of your laboratory records and results are confidential and shall not be disclosed without your written authorization.

Patient Attestation of Informed Consent:

My signature below indicates that I have received information about this test, **Celiac Disease GenotypR™**, and that I have read and understood the material in this document. I have been given a full opportunity to ask questions that I may have about the testing procedure and related issues. I agree to undergo this testing.

Patient Signature

Date

Signature of Parent/Guardian if Patient is a minor

Print Name of Parent/Guardian

For the Physician:

As the referring physician, I understand the benefits and limitations of this study and have requested that the above-named patient be tested. I attest to the fact that I have provided the patient with the information contained above and fully answered any questions. I believe that the patient understands the information and is voluntarily signing this informed consent.

Signature of Physician/Health Care Professional
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Print Name of Physician/Health Care Professional
Consent: Celiac Disease