

Celiac Disease Panel

Genetic Analysis Report



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PATIENT ORDERING PROVIDER

Example Organization

Lab ID: N8C9841

DOB: 01/01/72

Collection Date: 01/21/10

Test Date: 01/22/10

GENE MARKER	TEST RESULT	RISK ALLELE
DQA1	DQA1*04	NO
DQA1	DQA1*05	YES
DQB1	DQ2	YES
DQB1	DQ8	YES

The genetic risk for development of Celiac Disease based on this individual's reported HLA-DQA/DQB genotypes below is 1:7



VERY HIGH	HIGH	POPULATION	LOW	EXTREMELY LOW
1:7 1:10 1:12	1:24 1:26 1:35 1:89	1:100	1:210	1:1842 1:2518

BACKGROUND INFORMATION

Celiac disease (CD), non-celiac gluten sensitivity (NCGS) and wheat allergy (WA) are all distinct conditions whose primary treatment is avoidance of specific dietary components. Treatment for CD and NCGS is to remove gluten from the diet. Gluten is a protein naturally found in wheat, rye and barley, as well as in hybrids and products made from these grains. Treatment for wheat allergy is removal of all forms of wheat from the diet.

Celiac disease is an autoimmune disease that occurs in genetically predisposed individuals, in which the ingestion of gluten leads to an immune attack of the villi of the small intestines. Healthy villi are essential for the digestion and absorption of nutrients. Specific HLA-DQA1 and HLA-DQB1 genes represent the major genetic predisposition to Celiac that make assessment of the CD relative risk possible. It is important to note that although a positive test is indicative of genetic susceptibility, it does not necessarily mean the disease development. A negative test has a more significant value because gluten intolerance rarely occurs in the absence of HLA risk genes. The individuals who test negative for the HLA risk markers for Celiac, but continue to display the serious gastrointestinal symptoms after consuming a gluten-containing meal may have a condition known as non-celiac gluten sensitivity (NCGS). Such patients exhibit many of the same symptoms of celiac disease which are also resolved with the avoidance of gluten-containing foods. NCGS is distinct from Celiac and wheat allergy and it remains a diagnosis of exclusion as there is no standard approach to making a diagnosis.

Wheat allergy is an immune reaction to any of the hundreds of proteins in wheat. Symptoms of an allergy to wheat can include itching, hives, or anaphylaxis, a life-threatening reaction. When a person has a wheat allergy, one type of white blood cells, called B-cells, send out immunoglobulin E (IgE) antibodies to "attack" the wheat. At the same time, local tissues in the body send out natural chemical messengers to alert the rest of the body that lead to the observed WA symptoms. A person with a wheat allergy must avoid eating any form of wheat, but does not have trouble tolerating gluten from non-wheat sources. It is, however, possible for a person to be both allergic to wheat and have CD or NCGS.

SCIENTIFIC REFERENCES

- 1. Megiorni F et al. HLA-DQ and risk gradient for celiac disease. Human Immunology 2009 70: 55-59.
- 2. Megiorni F and A Pizzuti. HLA-DQA1 and HLA-DQB1 in Celiac disease predisposition: practical implications of the HLA molecular typing. J of Biomedical Science 2012. 19:88.
- 3. Abadie V et al. Integration of genetic and immunological insights into a model of celiac disease pathogenesis. Annu. Rev. Immunol. 2011. 29:493-525.
- 4. Kagnoff MF. Celiac disease: pathogenesis of a model immunogenetic disease. J Clin Invest 2007, 117:41-49.
- 5. Megiorni F, et al. HLA-DQ and susceptibility to celiac disease: evidence for gender differences and parent-of-origin effects. Am J Gastroenterol 2008. 103:997-1003
- 6. Karell K et al. HLA types in celiac disease patients not carrying the DQA1*05-DQB1*02(DQ2) heterodimer: results from the European genetics cluster of celiac disease. Hum Immunol 2003: 64:469-477.
- 7. Capannolo A et al. Non-Celiac Gluten Sensitivity among Patients Perceiving Gluten-Related Symptoms. Digestion. 2015; 92(1):8-13.
- 8. Elli L et al. Diagnosis of gluten related disorders: Celiac disease, wheat allergy and non-celiac gluten sensitivity. World J Gastroenterol. 2015; 21(23): 7110-7119.

This test detects only specific targeted genetic variations and there is a possibility that other genetic variants not detected by this test may be present. The DNA variants tested for in this report have been scientifically determined to be possible risk factors for the reported condition. The content of this report is provided for informational purposes only, not as a diagnostic tool. The report does not supersede the judgment of a qualified medical provider. This test is not a substitute for a comprehensive consideration of all factors that influence the maintenance of a healthy body. Genetic risk factors are not guarantees that you will develop a condition, and in many cases, the presence of a particular DNA variant may only play a minor role in your risk for disease, compared with environmental and lifestyle factors. This test is not FDA approved. The test's performance characteristics have been established and maintained by Kashi Clinical Laboratories under CLIA and CAP compliance.

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