



### PATIENT

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### ORDERING PROVIDER

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### LABORATORY INFORMATION

Lab ID: 170126034HLA  
Collection Date: 01/09/17  
Test Date: 01/26/17  
Report Date: 01/30/17

Celiac disease (CD) is a chronic gluten-intolerance that occurs in genetically predisposed individuals. The Kashi Celiac Panel tests two particular genes called DQA1 and DQB1, and each person has two copies of both genes, one from each parent. The test evaluates several different variations (alleles) of these genes to give an overall assessment of risk for developing celiac disease over a person's lifetime. Some of the alleles tested carry more risk than others, with some allele combinations having a higher risk of developing celiac disease and others having just a small risk. Over 40% of the population carries one or more genetic markers for celiac disease but only 1% of the population will actually develop celiac disease. Someone with several of the risk alleles will have increased risk for developing celiac disease.

GENE MARKER	TEST RESULT	RISK ALLELE	COMMENT
DQA1	DQA1*03	○	Extremely low risk allele except in combination with DQ8
DQA1	DQA1*05	◐	Medium risk allele
DQB1	DQ2	●	Significant risk allele
DQB1	DQ8	●	Significant risk allele

RISK ALLELE KEY: ○ No Risk ◐ Risk Allele Heterozygous ● Risk Allele Homozygous

### Overall Assessment of Risk for Developing Celiac Disease is 1:7



### BACKGROUND INFORMATION

The small intestine is lined with structures called villae which are part of the interface between nutrients in the small intestine, and the blood stream. The villae absorb nutrients which pass through into the blood stream to be distributed to cells throughout the body. The stomach and gut have lots of defense mechanisms to make sure that harmful substances do not pass through into the blood. Human leukocyte antigens (HLA) are proteins that help the body's immune system tell the difference between its own cells and foreign, harmful substances. For people with CD, the body's immune system is triggered by the presence of gluten, causing an inflammatory response which results in the aggression of the cells that form the lining of the gut. This immune response does not imply that the immune system is weakened and will not be able to respond appropriately to unwanted organisms. As long as gluten is consumed



however, the immune system over-responsiveness continues, and this leads to chronic inflammation of the mucosa lining of the small intestine, which frequently leads to deterioration of the villae that are so important for absorbing nutrients. It is common for individuals with CD to have deficiencies in key nutrients that impact the function of many cells in the body.

Celiac disease is associated with two specific genes called HLA DQA1 and HLA DQB1. There are several variations (alleles) in these genes that impact an individual's risk for developing CD. There is a category of genetic testing called Human Leucocyte Antigen testing, and this testing can be used to identify alleles, and thus a person's risk for developing CD. A report showing positive result for several of the alleles does not mean the person will definitely get CD, it is a risk assessment for the likelihood of developing the disease. The genes are not standalone and the way they work creates interconnection between them, some of which scientists understand, while others are still a mystery.

Allele names associated with risk: DQA1\*05, DQ2, DQ8

CD occurs in approximately 1 in 100 people and is more likely to occur in females than males by about 2:1. CD is more frequent in at-risk groups, such as first-degree relatives of those with specific genetic syndromes such as Down, Turner, Williams, or autoimmune conditions such as type 1 diabetes, thyroiditis and multiple sclerosis.

### GLOSSARY

**Allele** - Any of the alternative forms of a gene that may occur at a given locus.

**Antigen** - A molecule capable of promoting an immune response.

**HLA Typing** - A specific type of genetic testing used to evaluate part of the immune system.

**Leukocytes** - Cells of the immune system that are involved in protecting against infectious disease and foreign invaders.

**Locus** - A particular location of a gene or genetic marker.

### SCIENTIFIC REFERENCES

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*This test was developed and its performance characteristics determined by Kashi Clinical Laboratories. It has not been cleared or approved by the FDA. The laboratory is regulated under CLIA as qualified to perform high-complexity testing. This test is used for clinical purposes. It should not be regarded as investigational or for research.*

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