



CELIAC DISEASE GENETIC TESTING

Healthcare Professional Information

The Celiac Panel

Celiac disease (CD) is a chronic gluten-intolerance that occurs in genetically predisposed individuals.¹ The Celiac Panel tests two particular genes called DQA1 and DQB1. Each person has two copies of both genes, one from each parent.² 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.³ 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.^{4,5} Someone with several of the risk alleles will have increased risk for developing celiac disease.⁶

Common Symptoms: Chronic burping, heat burn/reflux, gas/bloating, constipation and or diarrhea.

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 many 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.

References:

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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. Karelk 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.



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