

## Level 1

### Genetics (Gene Testing)

#### KEY POINTS:

- Most people with celiac disease (CD) have the genes for HLA-DQ2 or DQ8. These genes are made of DNA. Genes are the part of our cells that tell our bodies how to grow and work. Genes are passed from parent to child, so you usually share the same genes with your relatives.
- However, these genes are also present in 30–40% of the western Caucasian population. Only 3% of individuals with these genes develop CD.<sup>1</sup>
- CD is caused by both genetic and environmental triggers. Just because you have the right genes for CD does NOT mean you will get it. In fact, most people with the HLA-DQ2 and DQ8 genes do NOT get CD. Scientists are still studying what other things cause people to develop CD.<sup>2</sup>
- Genetic testing can tell us whether you have the HLA-DQ2 or DQ8 genes. If you have one or both of them, it does NOT mean you have CD. You will need further testing to find out if you have CD. If you do NOT have them, you most likely do NOT have CD.<sup>2</sup>
- If one person in your family has the genes for CD, other people in the family probably have them, too.
- However, HLA-DQ2 / DQ8 testing is not used routinely in the initial diagnosis of CD.<sup>2</sup>
- In selected clinical situations, HLA-DQ2/DQ8 is used to effectively rule out CD.<sup>3</sup>

#### TAKE HOME MESSAGES:

1. The vast majority of patients with CD have the HLA-DQ2 or DQ8 gene. Genetic testing for CD will tell you if you have them.
2. If you have the DQ2 or DQ8 genes, it does NOT mean that you have CD. It does mean that further testing is needed.
3. If you do not have these genes, you probably do not have CD.
4. These tests are not used routinely for initial diagnosis of CD. In select clinical situations, the celiac gene test can effectively rule out CD.

#### References:

1. Ludvigsson JF, Bai JC, Biagi F, et al. Diagnosis and management of adult coeliac disease: guidelines from the British Society of Gastroenterology. *Gut* 2014;63:1210–1228.

2. Pietzak, M. Genetic Testing in Celiac Disease. In *Real Life with Celiac Disease: Troubleshooting and Thriving Gluten-Free*. Dennis M, Leffler D, eds. AGA Press. Bethesda, MD, 2010.
3. Rubio-Tapia A, Hill ID, Kelly CP, Calderwood AH, Murray JA, ACG Clinical Guidelines: Diagnosis and Management of celiac disease. 2013, Am J Gastroenterol 108:656-676.

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