CeliacDx™
5 Mason Irvine, CA 92618

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CAP 9145700
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Name
Date of Birth
CeliacDx Kit ID
Lab Accession #

Clinical History

Intestinal and extra-intestinal symptoms. Positive family history of celiac disease. t-Transglutaminase (tTG) IgA antibody testing is positive. Biopsy is negative. Celiac genetic risk testing is positive.

Four out of Five of the following criteria are used for a definitive celiac disease diagnosis:

1. Positive Celiac symptoms (SX)
2. Positive Celiac Genetics
3. Positive tTg Antibodies (AB)
4. Positive Biopsy (BX)
5. Response to gluten free diet (GFD).

Catassi and Fasano. The American Journal of Medicine (2010) 123, 691-693

Four out of Five Rule for Celiac Disease Diagnosis

THREE out of FIVE criteria are met:
Positive Symptoms, Genetics, Antibodies

Diagnostic Reference Table for Celiac Disease Positive Results

Celiac Disease Classification	Antibodies	Biopsy	Genetics
Celiac Disease	Positive	Positive	Positive
Potential Celiac Disease	Positive	Negative	Positive
Seronegative Celiac Disease	Negative	Positive	Positive
Negative Celiac Disease	Negative	Negative	Negative

Reference: Celiac Disease: A Comprehensive Current Review. CAIO et al. BMC Medicine (2019) 17:142 https://doi.org/10.1186/s12916-019-1380-z

Comprehensive Assessment

Positive genetics, positive symptoms, and positive antibodies consistent with potential celiac disease.

Recommendation

Share these results with your heath care provider or our affiliate Functional Medicine provider Root Cause Medical Clinics before making any lifestyle or dietary changes. Telemedicine available through Root Cause. Call 727-335-0400 or set up an online consultation at https://rootcausemedicalclinics.com.

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Your Genetics Report At-A-Glance

Your Result

1 Celiac Risk Variant Was Detected

Your GlutenID is:

DQ8

What does this mean?
You have a genetic risk for celiac disease. Symptoms of celiac disease can be triggered when a person with a genetic risk eats gluten in their diet.

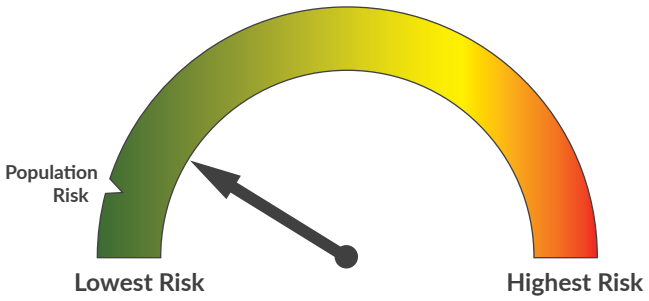
This result does not mean you have or will develop celiac disease. Many people who have a genetic risk never develop the condition.

Your Risk

Slightly Increased Risk

Based on DNA testing, you have a slightly increased risk for celiac disease.

If you have symptoms of celiac disease or a family history, your actual risk may be higher.



Important Next Steps

Talk to your healthcare provider
Share your result with them. They can discuss next steps for you based on your health and family history. Your healthcare provider may recommend other tests for celiac disease. For people who have celiac disease, removing gluten from their diet is usually an effective treatment. **Talk to your healthcare provider before making any changes to your diet or lifestyle.**

Talk to your family
Celiac disease runs in families. There is a chance your parents, siblings, and children also have the DQ2(cis) variant. Your result might be important for their health too.

Limitations of the Test

Important things to know

It **does not diagnose** celiac disease, gluten or wheat-related sensitivities, or any other condition.
It **cannot predict** if someone will develop celiac disease in the future.
It **does not replace** a visit with your healthcare provider.
It **does not detect** all possible genetic risk factors for celiac disease.
It is based on **current scientific knowledge**. This will continue to improve over time.

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Scientific Details

Your DNA Results

Haplotype	Other Names for These Haplotypes	Tag SNP	Variant Genotype	Your Genotype	Number of Copies
HLA-DQ2	DQ2(cis) DQ2.5	rs2187668	T	C / C	0
HLA-DQ8	**DQ8**	**rs7454108**	**C**	**T / C**	**1**
HLA-DQ2.2	half-DQ2	rs7775228	C	T / T	0
HLA-DQ7	half-DQ2	rs4639334	A	G / G	0

Note: Two additional SNPs rs2395182 and rs4713586 are used for confirmation of DQ2. For more scientific information see GlutenID Technical Details.

What is the GlutenID Test Looking For?

The GlutenID test detects the presence or absence of celiac risk haplotype variants. It looks for markers (or tag SNPs) associated with celiac risk haplotypes located close to the HLA-DQA1 and HLA-DQB1 genes.

Four specific haplotypes are associated with a risk for celiac disease, commonly called DQ2, half-DQ2, DQ7, and DQ8

About these variants:

- 95% of people with celiac disease have at least one copy of DQ2 or DQ8.
- These variants are seen in people from many ethnicities.
- They are common; 30% of people in the general population have at least one copy of DQ2 or DQ8.
- However, only 3% of people with at least one copy of DQ2 or DQ8 will develop celiac disease—most people with DQ2 or DQ8 will not develop celiac disease.

Your Risk Estimate

Risk estimates are based on clinical studies. This table summarizes the estimated genetic risk and risk category associated with each GlutenID result.

You have a slightly increased risk to develop celiac disease compared to people with no genetic risk.

GlutenID	Estimated Risk	Risk Category
DQ2+DQ8	1 in 7	Increased Risk
DQ2+DQ2	1 in 7	Increased Risk
DQ2+half-DQ2	1 in 10	Increased Risk
DQ8+DQ8	1 in 12	Increased Risk
DQ8+half-DQ2	1 in 24	Slightly Increased Risk
half-DQ2+half-DQ2	1 in 26	Slightly Increased Risk
DQ2+DQ7	1 in 35	Slightly Increased Risk
DQ2(cis)	1 in 35	Slightly Increased Risk
DQ2(trans)	1 in 35	Slightly Increased Risk
DQ8+DQ7	1 in 89	Slightly Increased Risk
DQ8	**1 in 89**	**Slightly Increased Risk**
half-DQ2	1 in 210	Slightly Increased Risk
DQ7+DQ7	1 in 1842	Slightly Increased Risk
DQ7	1 in 1842	Slightly Increased Risk
Non-Celiac Genetics	1 in 2518	Not Likely At Risk

Click here and more information on the GlutenID risk categories.

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Peer-Reviewed Journal Articles

1 Koskinen L et al. (2009). "Cost-effective HLA typing with tagging SNPs predicts celiac disease risk haplotypes in the Finnish, Hungarian, and Italian populations." Immunogenetics. 61(4):247 56. ncbi.nlm.nih.gov/pubmed/19255754

2 Liu E et al. (2017) "High incidence of celiac disease in a long-term study of adolescents with susceptibility genotypes." Gastroenterol. 152:1329-1336. pubmed.ncbi.nlm.nih.gov/28188747/

3 Megiorni F et al. (2009) "HLA-DQ and risk gradient for celiac disease." Hum Immunol 70:55-59. pubmed.ncbi.nlm.nih.gov/19027045/

4 Megiorni F et al. (2012). "HLA-DQA1 and HLA-DQB1 in celiac disease predisposition: practical implications of the HLA molecular typing." J Biomed Sci 19:88. pubmed.ncbi.nlm.nih.gov/23050549/

5 Monsuur AJ et al. (2008). "Effective detection of human leukocyte antigen risk alleles in celiac disease using tag single nucleotide polymorphisms." PLoS One. 3(5):e2270. ncbi.nlm.nih.gov/pubmed/18509540

6 Nellikkai S et al. (2019) "High prevalence of celiac disease among screened first-degree relatives." Mayo Clin Proc.94(9):1807-1813. pubmed.ncbi.nlm.nih.gov/31447136/

7 Singh P et al. (2018). "Global prevalence of celiac disease: systematic review and meta-analysis." Clin Gastroenterol and Hepatol. 16:823-836. pubmed.ncbi.nlm.nih.gov/29551598/

8 Singh P et al. (2015). "Risk of celiac disease in the First- and Second-Degree Relatives of Patients with celiac disease: A Systematic Review and Meta-Analysis." Am J Gastroenterol. 110(11):1539-48. ncbi.nlm.nih.gov/pubmed/26416192

9 Taylor AK et al. (2008). "Celiac disease." [Updated 2019 Jan 31. ncbi.nlm.nih.gov/pubmed/20301720

10 Tinto N et al. (2015) "High frequency of haplotype HLA-DQ7 in celiac disease patients from South Italy: retrospective evaluation of 5,535 subjects at risk of celiac disease." PLoS ONE 10(9); e0138324 pubmed.ncbi.nlm.nih.gov/26398634/

11 Guidi A et al. (2018) "Celiac disease testing." College of American Pathologists (CAP) member resources.

12 Pietzak M et al. (2009) "Stratifying risk for celiac disease in a large at-risk United States population by using HLA alleles." Clin Gastroenterol Hepatol. 7(9):966-971. pubmed.ncbi.nlm.nih.gov/19500688/

13 Rubio-Tapai A et al. (2013) ACG clinical guidelines: diagnosis and management of celiac disease." Am J Gastro. 108:656-676. pubmed.ncbi.nlm.nih.gov/23609613/

14 Farmer, G et al (2020). "Recommendations for designing genetic test reports to be understood by patients and non-specialists." Eur J Hum Genet. 28: 885–895. https://doi.org/10.1038/s41431-020-0579-y

LabCorp
Laboratory Corporation of America

Testmaster Testing
3060 S Church Street
Burlington, NC 27215

Phone: **336-436-2762**

Specimen Number	Patient ID	Control Number	Account Number	Account Phone Number	Route
322-988-3212-0			90000999	336-436-8645	00

Patient Last Name	Account Address
SAMPLE REPORT	LabCorp Test Master Test Account 3060 South Church Street Burlington NC 27215

Patient First Name	Patient Middle Name
164640	

Patient SS#	Patient Phone	Total Volume

Age (Y/M/D)	Date of Birth	Sex	Fasting
23	01/01/01	M	

Patient Address	Additional Information
	ABNORMAL REPORT

Date and Time Collected	Date Entered	Date and Time Reported	Physician Name	NPI	Physician ID
08/31/23 00:00	8/31/23				

Tests Ordered
t-Transglutaminase (tTG) IgA

TESTS	RESULT	FLAG	UNITS	REFERENCE INTERVAL	LAB
t-Transglutaminase (tTG) IgA	**10**	**High**	U/mL	0 - 3	01
			Negative	0 - 3	
			Weak Positive	4 - 10	
			Positive	>10	

Tissue Transglutaminase (tTG) has been identified as the endomysial antigen. Studies have demonstrated that endomysial IgA antibodies have over 99% specificity for gluten sensitive enteropathy.

01 BN LabCorp Burlington Dir: William F Hancock, MD
1447 York Court, Burlington, NC 27215-3361
For inquiries, the physician may contact **Branch: 800-222-7566 Lab: 336-436-2762**

Gastro Pathology Lab Pathology Report

Patient: Any Patient
Address: Anywhere, USA
Phone: (123) 456-7891

Age: 23
Date of Birth: 01/01/23

Client: Endoscopy Center
Doctor: D. Doctor M.D.

Accession ID:123456789

Collection Date: 8/31/2023
Date Rec: 8/31/2023

FINAL DIAGNOSIS

1. **Duodenum, biopsies:**
   - **Benign small bowel showing normal villous architecture with a nonspecific diffuse increase of mucosal lymphocytes confirmed by CD3 immunostain.**
   - **No parasitic organisms identified.**
   - **Please see microscopic.**

2. **Next stomach, antrum, biopsies:**
   - **Inactive chronic antral gastritis.**
   - **Negative for intestinal metaplasia.**
   - **Negative for dysplasia or malignancy.**
   - **No evidence of Helicobacter on routine stain or by immunostain.**

3. **Stomach, body, biopsies:**
   - **Mild inactive chronic gastritis.**
   - **Negative for intestinal metaplasia.**
   - **Negative for dysplasia or malignancy.**
   - **No evidence of Helicobacter on routine stain or by immunostain.**

4. **Esophagus, distal, biopsies:**
   - **Hyperplastic squamous epithelium representing reflux.**
   - **With gastric mucosa shoiwing inactive chronic gastritis.**
   - **Negative for Barrett's esophagus.**
   - **No evidence of Helicobacter.**
   - **Negative for eosinophilic esophagitis, dysplasia, or malignancy.**

Electronically Signed out by:
D. **Pathologist** M.D.
Board certified in Anatomic & Clinical Pathology

Date: Time:
9/5/2023 9:44 AM Patient ID: 123456

CLINICAL DESCRIPTION

Preop diagnosis: **Family history of celiac disease**, abdominal pain

GROSS DESCRIPTION

1. Duodenum, rule out celiac sprue (CD3). 2 biopsies. TE A1.
2. Antrum, rule out H. pylori (IHC). 2 biopsies. TE B1.
3. Body, rule out H. pylori (IHC). 2 biopsies. TE C1.
4. Distal esophagus, rule out Barrett's (ABPASH). 2 biopsies. TE D1. (jt) 08/31/2023

MICROSCOPIC DESCRIPTION

Microscopic examination performed. See diagnosis.

Specimen 1: After reviewing the H&E stained slide which demonstrated some mucosal lymphocytosis, immunostain for CD3 was ordered to accurately assess the number of lymphocytes/100 enterocytes. This stain shows a diffuse increase of intraepithelial lymphocytes but preserved villous architecture. The number of CD3 positive lymphocytes/100 enterocytes is > 30 but < 40 and thus sprue is not totally excluded. Conditions that might give this histologic appearance include early or partially treated gluten and nongluten food allergies, autoimmune disorders, bacterial overgrowth, Helicobacter pylori gastritis, and as a response to medication such as NSAIDs. This may also be a variant of normal. Clinical correlation is recommended.