Warde Medical Laboratory

LABORATORY REPORT

Example Client, XYZ123
1234 Warde Road
Ann Arbor MI 48108

EXAMPLE, REPORT W
WX0000003827 M 07/08/1978 45 Y

Referral Testing

Collected: 08/30/2023 14:07 Received: 08/30/2023 14:07

Test Name	Result	Flag	Ref-Ranges	Units	Site
Celiac Disease HLA-DQ Genotyping					
HLA-DQA1, Allele 1	01:02				ARRL
Performed By: UUH Histocompatibility and Immunogenetic 417 Wakara Way Suite 3220 Salt Lake City, UT 84108 CLIA Number: 46D0523979					
HLA-DQA1, Allele 2	03:01				ARRL
Performed By: UUH Histocompatibility and Immunogenetic 417 Wakara Way Suite 3220 Salt Lake City, UT 84108 CLIA Number: 46D0523979					
HLA-DQB1, Allele 1	03:02				ARRL
Performed By: UUH Histocompatibility and Immunogenetic 417 Wakara Way Suite 3220 Salt Lake City, UT 84108 CLIA Number: 46D0523979					
HLA-DQB1, Allele 2	06:04				ARRL
Performed By: UUH Histocompatibility and Immunogenetic 417 Wakara Way Suite 3220 Salt Lake City, UT 84108 CLIA Number: 46D0523979					
Celiac HLA Interpretation	See Note				ARRL

Positive for HLA-DQ8 (DQB1*03:02), one copy.
Interpretation: HLA-DQ8 was detected. At least one copy of this allele is observed in approximately 5-10 percent of individuals with celiac disease and 15-20 percent of the general population. This result is supportive of a clinical diagnosis of celiac disease, but does not by itself establish a diagnosis. If this individual is an asymptomatic relative of an affected individual, celiac disease-associated antibody testing should be performed at

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three to five year intervals. Medical screening and management of this individual should rely on clinical findings.
Performed By: UUH Histocompatibility and Immunogenetic
417 Wakara Way
Suite 3220
Salt Lake City, UT 84108
CLIA Number: 46D0523979
BACKGROUND INFORMATION: Celiac Disease HLA-DQ Genotyping

CHARACTERISTICS: Celiac disease is a systemic autoimmune disease of the gastrointestinal system caused by exposure to cereal gluten in genetically susceptible individuals.

INCIDENCE: On average, 1 in 133 individuals in the United States is affected.

INHERITANCE: Multifactorial.

CAUSE: The presence of either HLA-DQ2 or the HLA-DQ8 alleles in combination with dietary gluten.

CLINICAL SENSITIVITY: greater than 99 percent.

METHODOLOGY: Polymerase Chain Reaction/Massively Parallel Sequencing, or Polymerase Chain Reaction/Sequence-Specific Oligonucleotide Probe Hybridization.

ANALYTICAL SENSITIVITY AND SPECIFICITY: greater than 99 percent.

LIMITATIONS: Rare diagnostic errors may occur due to primer site mutations. Other genetic and nongenetic factors that influence celiac disease are not evaluated. In cases where an HLA allele cannot be resolved unambiguously, the allele assignment will be reported as the most common, based on allele frequencies from the common, intermediate and well-documented alleles catalogue version 3.0.0 (Hurley CK et al, 2020).

ALLELES TESTED: HLA-DQA1 and HLA-DQB1 alleles.

Most celiac disease patients (approximately 90 percent) carry HLA-DQ2.5 heterodimers encoded by HLA-DQA1*05 and HLA-DQB1*02 alleles. The remaining 5-10 percent of the patients carry HLA-DQ8, encoded by HLA-DQB1*03:02 allele, most commonly in combination with HLA-DQA1*03 alleles. A minority of patients negative for the above genotypes may

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carry HLA-DQB1*02 but without the DQA1*05 alpha chain, most commonly with DQA1*02. The presence of the DQB1*02 allele in combination with either DQ2.5 or DQ8 may further increase celiac disease risk.

Stratified overall genetic risk for patients carrying the celiac disease-associated HLA-DQ genotypes:

Genotype	Risk*
DQ2.5 homozygous	Very High (greater than 1:10)
DQ2.5 + DQB1*02	Very High (greater than 1:10)
DQ2.5 + DQ8	High (greater than 1:20)
DQ8 homozygous	High (greater than 1:20)
DQ8 + DQB1*02 (without DQA1*05)	Intermediate (greater than 1:50)
DQ2.5 heterozygous	Intermediate (greater than 1:50)
DQ8 heterozygous	At risk (greater than 1:100)
Population risk for unknown genotype	1:100
DQB1*02 (without DQA1*05)	Low
DQA1*05 (without DQB1*02)	Minimal
Negative for DQ2 and DQ8	Not at risk

* Risk is provided from the references below, and defined according to HLA allele combinations, considering a disease prevalence of 1:100. However, these alleles are common in the general population and the majority of individuals positive for celiac-associated alleles do not develop the disease. Detection of these alleles can support a clinical diagnosis but should not be interpreted as diagnostic of celiac disease.

References:

1. Megiorni F, Mora B, Bonamico M, et al. HLA-DQ and risk gradient for celiac disease. Human Immunology. 2009;70:55-59.

2. Pietzak MM, Schofield TC, McGinnis MJ, et al. Stratifying risk for celiac disease in a large at-risk United States population by using HLA alleles. Clinical Gastroenterology and Hepatology. 2009;7:966-971.

3. Almeida LM, Gandolfi L, Pratesi R, et al. Presence of DQ2.2 associated with DQ2.5 increases the risk for celiac disease. Autoimmune Diseases, 2016. 2016:5409653.

4. Vader W, Stepniak D, Kooy Y, et al. The HLA-DQ2 gene dose effect in celiac disease is directly related to the magnitude and breadth of gluten-specific T cell responses. PNAS. 2003;100:12390-12395.

DISCLAIMER INFORMATION:

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This test was developed and its performance characteristics determined by the Histocompatibility& Immunogenetics laboratory at the University of Utah Health. It has not been cleared or approved by the US Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary. This test is used for clinical purposes. It should not be regarded as investigational or for research. Histocompatibility& Immunogenetics laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA-88) as qualified to perform high complexity clinical laboratory testing.
Performed at: Histocompatibility & Immunogenetics Laboratory, University of Utah Health, 417 Wakara Way, Suite 3220, Salt Lake City, UT 84108.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

Performing Site:
ARRL: ARUP REFERENCE LAB 500 Chipeta Way Salt Lake City UT 841081221

Reported Date: 2023.08.30 14:07 CDHDG

LAB: L - LOW, H - HIGH, AB - ABNORMAL, C - CRITICAL, . - NOT TESTED

F230000038
WX0000003827
Printed D&T: 08/30/23 14:07

Ordered By: KAJAL SITWALA, MD, PhD
WX00000000002365

Kajal V. Sitwala, MD, PhD - Medical Director
Form: MM RL1