Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB	9/18/2000
Sex:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

Celiac Disease HLA-DQ Genotyping

ARUP test code 3004445

HLA-DQA1, Allele 1	01 Performed By:
HLA-DQA1, Allele 2	01:02 Performed By:
HLA-DQB1, Allele 1	05:02 Performed By:
HLA-DQB1, Allele 2	05:01 Performed By:
Celiac HLA Interpretation	See Note Negative for HLA-DQA1*05, HLA-DQB1*02 and HLA-DQB1*03:02. Interpretation: None of the alleles associated with celiac disease were detected. Since over 99 percent of affected individuals have at least one copy of a risk allele, this individual is not expected to be at risk for developing celiac disease. Performed By:

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.

H=High, L=Low, *=Abnormal, C=Critical

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 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:

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  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
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* 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:
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:

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
HLA-DQA1, Allele 1	22-125-138776	5/5/2022 8:00:00 AM	5/6/2022 7:09:56 AM	5/6/2022 7:15:00 AM
HLA-DQA1, Allele 2	22-125-138776	5/5/2022 8:00:00 AM	5/6/2022 7:09:56 AM	5/6/2022 7:15:00 AM
HLA-DQB1, Allele 1	22-125-138776	5/5/2022 8:00:00 AM	5/6/2022 7:09:56 AM	5/6/2022 7:15:00 AM
HLA-DQB1, Allele 2	22-125-138776	5/5/2022 8:00:00 AM	5/6/2022 7:09:56 AM	5/6/2022 7:15:00 AM
Celiac HLA Interpretation	22-125-138776	5/5/2022 8:00:00 AM	5/6/2022 7:09:56 AM	5/6/2022 7:15:00 AM

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