

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: [REDACTED]

Gender: [REDACTED]

Patient Identifiers: 01234567890ABCD, 012345

Visit Number (FIN): 01234567890ABCD

Collection Date: 00/00/0000 00:00

Celiac Disease (HLA-DQ2, and HLA-DQ8) Genotyping

ARUP test code 2005018

HLA Celiac Specimen whole Blood

Celiac (HLA-DQA1*05) **Positive** *

Celiac (HLA-DQB1*02) **Positive** *

Celiac (HLA-DQ8) Negative

Celiac HLA Interpretation See Note

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.

Result: Positive for HLA-DQA*05 and HLA-DQB*02
Interpretation: The HLA-DQA*05 and HLA-DQB*02 alleles were detected. This genotype is observed in approximately 90 percent of individuals with celiac disease and 20-30 percent of the general population. This result is supportive of a clinical diagnosis of celiac disease, but by itself does not establish a diagnosis. If this individual is an asymptomatic relative of an affected individual, celiac disease-associated antibody testing should be performed at three to five year intervals. Medical screening and management of this individual should rely on clinical findings.

This result has been reviewed and approved by Julio Delgado, M.D., M.S.

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

BACKGROUND INFORMATION: Celiac Disease (HLA-DQ2 and HLA-DQ8) Genotyping

CHARACTERISTICS: Celiac disease is a systemic autoimmune disorder that may be associated with gastrointestinal symptoms including: diarrhea, weight loss, anorexia, lactose intolerance, and abdominal distention and discomfort. Non-gastrointestinal characteristics are highly variable and include: chronic fatigue, joint pain/inflammation, migraines, epilepsy, depression, attention deficit disorder, iron-deficiency anemia, vitamin deficiency, osteoporosis/osteopenia, short stature, delayed puberty, dental enamel defects, infertility, recurrent fetal loss, and dermatitis herpetiformis.
INCIDENCE: One in 133 individuals in the US is affected.
INHERITANCE: Multifactorial.
CAUSE: The presence of either the HLA-DQ2 or the HLA-DQ8 allele in combination with dietary gluten.
ALLELES TESTED: HLA-DQ2 (encoded by HLA-DQA1*05 and HLA-DQB1*02); and HLA-DQ8 (encoded by HLA-DQB1*03:02).
CLINICAL SENSITIVITY AND SPECIFICITY: Approximately 100 percent and 3 percent, respectively.
METHODOLOGY: PCR with melting curve analysis.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
LIMITATIONS: Rare diagnostic errors may occur due to primer site mutations. Copy number of each detected allele will not be determined. Alleles other than HLA-DQ2 and HLA-DQ8 will not be identified. Other genetic and non-genetic factors that influence celiac disease are not evaluated.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
HLA Celiac Specimen	19-060-129083	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Celiac (HLA-DQA1*05)	19-060-129083	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Celiac (HLA-DQB1*02)	19-060-129083	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Celiac (HLA-DQ8)	19-060-129083	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Celiac HLA Interpretation	19-060-129083	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

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