



TEST ID: Gut Health Profile (GHP)
DESCRIPTION: HLA Typing for Celiac Disease

COMPONENTS

HLA-DQ2.5, HLA-DQ8

SUB-COMPONENTS

(DQ2.5) DQA1*05 plus DQB1*02
(DQ8) DQA1*03 plus DQB1*0302

CLINICAL USE

To determine genetic susceptibility to celiac disease (CD)
To rule out CD diagnosis

*Certain human leukocyte antigen (HLA) alleles in the DQ region have been linked to a predisposition to CD. Approximately 95% of patients with CD have the HLA-DQ2 heterodimer determined by the DQA1*05 and DQB1*02 alleles, while close to 5% have the HLA-DQ8 heterodimer determined by the DQA1*03 and DQB1*0302 alleles.¹ Since 25% to 40% of the United States population has either DQ2 or DQ8, the presence of either is not indicative of CD.¹ Thus, the primary use of HLA-DQ typing is to rule out CD and determine the risk of susceptibility for CD.*

SPECIMEN INFORMATION

COLLECTION

Whole blood in sodium citrate tube. (Use blue top tube included with collection kit)

SPECIMEN STABILITY

Stable at room temperature for 1 week.

REJECTION CRITERIA

Blood sample is over 1 week old; grossly hemolyzed, icteric, and lipemic specimens; improper anticoagulant used.

METHOD

Real-time SSP-PCR (Polymerase Chain Reaction using Sequence Specific Primers) followed by melt curve analysis

RESULT INTERPRETATION

A negative result for both HLA-DQ2 and HLA-DQ8 practically dismisses the diagnosis of CD and indicates an extremely low risk for subsequent development of the disease ($\geq 95\%$ negative predictive value). More than 97% of patients with celiac disease have either HLA-DQ2.5 or HLA-DQ8 genotype. Presence of either or both HLA-DQ2.5 and HLA-DQ8 does not mean CD will occur. In fact, the vast majority of people with the genes never develop CD. Having the genes may impact the risk of someday developing CD. On the other hand, if the genes are not present, the odds of developing CD are almost non-existent.

CPT CODES

HLA-DQA1*05	81377
HLA-DQB1*02	81377
HLA-DQA1*03	81377
HLA-DQB1*0302	81383

REFERENCES

1. Megiorni F, Mora B, Bonamico M, Barbato M, Nenna R, et al: HLA-DQ and risk gradient for celiac disease. Hum Immunol 2009, 70:55-59.