

## Patient Information

Name: PATIENT II, PRETEND

Date of Birth: 11/04/1977	Gender: F	Lab ID: 68220
Date Received: 02/11/2010	Date Collected:	Date Reported: 01/17/2017
Physician: Sample Physician	Clinic ID: 10804	

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## Celiac Disease Genetic Markers



\*These tests were performed using Polymerase Chain Reaction with Sequence Specific Primers (SSP-PCR) Technique.

## Genetic Markers - HLA-DQ Typing\*

HLA-DQ2.5	Positive
<b>DQA1*05</b>	Positive
<b>DQB1*02</b>	Positive

HLA-DQ8	Negative
<b>DQA1*03</b>	Negative
<b>DQB1*0302</b>	Positive

## HLA-DQ Typing Commentary

**The Risk for Celiac Disease Risk between 1:10 AND 1:35 (1)**

Patient has one of the HLA-DQ variants associated with Celiac Disease. More than 90% of Celiac patients carry either HLA-DQ2.5 or HLA-DQ8 or both. However, since almost 30% of general U.S population carry these variants, the presence of these are not an indication of Celiac Disease but only reflect a genetic predisposition for the disease.

(1) Megiomi 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.

(2) Megiomi F, Pizzuti, A. HLA-DQA1: HLA-DQB1 in Celiac Disease predisposition: practical implications of the HLA molecular typing.

## Crohn's Genetic Markers\*

<b>ATG16L1 (T300A)</b>	Homozygous Negative	Genes inherited from both parents do not have this mutation (homozygous negative genotype).
<b>NOD2 (L1007sinsC)</b>	Homozygous Negative	Genes inherited from both parents do not have this mutation (homozygous negative genotype).
<b>NOD2 (R702W)</b>	Homozygous Negative	Genes inherited from both parents do not have this mutation (homozygous negative genotype).
<b>NOD2 (G908R)</b>	Homozygous Negative	Genes inherited from both parents do not have this mutation (homozygous negative genotype).

## Crohn's Comments

Although there is an absence of the mutations, the etiologies of inflammatory bowel disease (IBD) and Crohn's disease are highly complex and they can occur without a mutation at the investigated sites.

(1) Aditya Murthy and Menno van Lookeren Campagne: Understanding Crohn's diseases through genetics. *Cell Cycle* 13:18, 2803-2804; September 15, 2014.

(2) Aditya Murthy et al: A Crohn's disease variant in Atg16l1 enhances its degradation by caspase 3. *Nature*, Vol 506, 27 February 2014.

(3) Denie K. Bohnen et al: Crohn's Disease-Associated NOD2 Variants Share a Signaling Defect in Response to Lipopolysaccharide and Peptidoglycan. *Gastroenterology* 2003;124:140-146.

\* This test was developed and its performance characteristics determined by Cell Science Systems. It has not been cleared or approved by the U.S. Food and Drug Administration.

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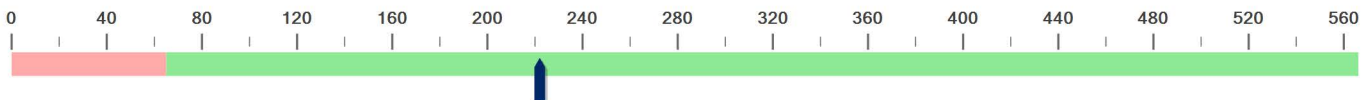
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## Serologic Markers

## Total IgA

222

Reference Range (age/gender based) :  
(65 - 421 mg/dL)

## Personalized Commentary

	NEGATIVE < 20.1 units	WEAK POSITIVE 20.1 - 30 units	MODERATE TO STRONG POSITIVE > 30 units	REMARKS
Tissue transglutaminase (tTg) IgA	5			
Tissue transglutaminase (tTg) IgG	3			
Deamidated gliadin peptide (DGP) IgA	11			
Deamidated gliadin peptide (DGP) IgG	14			

	NEGATIVE < 20.1 units	EQUIVOCAL 20.1 - 24.9 units	POSITIVE > 25 units	REMARKS
Anti-Saccharomyces cerevisiae Antibodies (ASCA) IgA	13.4			
Anti-Saccharomyces cerevisiae Antibodies (ASCA) IgG	3.5			

## Antibody Markers Commentary

A finding of tissue transglutaminase (tTG) IgA antibodies may be indicative for Celiac Disease. For patients with normal total IgA levels and negative tTG IgA antibodies results, an indication of Celiac Disease is very unlikely. However, it is important to remember that a certain percentage of patients with Celiac Disease may be seronegative. If the testing for tTG IgA is negative, but Celiac Disease is still suspected based on clinical presentation or even a strong family history, looking to the results of the DGP-IgA antibody test and the HLA DQ2.5/DQ8 genetic typing would be appropriate

A Gluten Restricted Diet prior to testing may affect the antibody Result.

**Patient Information**

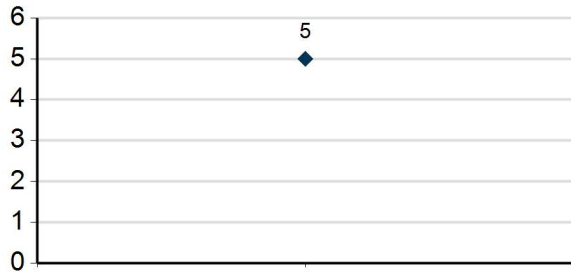
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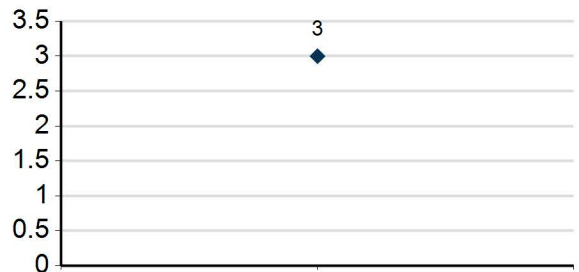
**Patient Historic Comparison**

68220							
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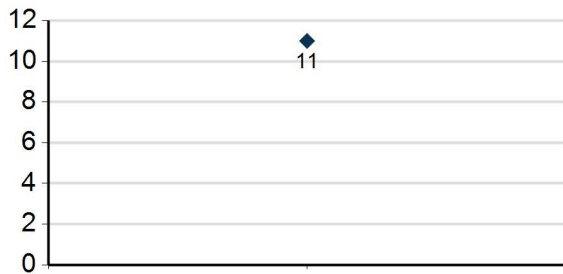
**tTg IgA**



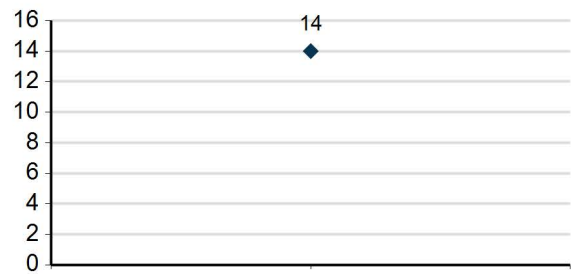
**tTg IgG**



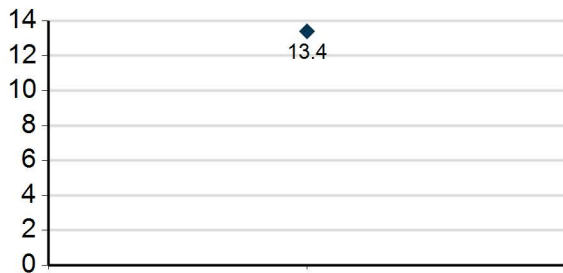
**DGP IgA**



**DGP IgG**



**ASCA IgA**



**ASCA IgG**

