

TEST REPORT  
Real Time PCR Genotyping by buccal swabs

TEST REPORT No \_\_\_\_\_  
Applicant \_\_\_\_\_  
Name and Surname \_\_\_\_\_  
Patient Date of birth \_\_\_\_\_  
Sample collection date \_\_\_\_\_  
Sample receipt date \_\_\_\_\_  
Analysis date \_\_\_\_\_  
Analysis code HLA molecular typing kit XeliGen XL

**RISK OF CELIAC DISEASE**

Alleles tested::

DQA1\*01, DQA1\*02:01, DQA1\*03, DQA1\*05, DQA1\*06, DQB1\*02, DQB1\*03:01, DQB1\*03:02, DQB1\*03:03, DQB1\*03:04, DQB1\*03:05, DQB1\*04, DRB1\*03, DRB1\*04, DRB1\*07, DRB1\*11

Genotype DQ	Genotype DR	Haplotypes Status	Status DQB1*02	Susceptibility
DQ2	DR3 (rari DR11, DR13)	DQA1*01 - DQB1*02	HOMOZYGOUS	
DQ2	DR3 (rari DR11, DR13)	DQA1*05 - DQB1*02		

Presence of heterodimer DQ2 (DQA1*05, DQB1*02)	YES in CIS
Presence of DQ8 (DQB1*0302)	NO

The presence of at least one of these conditions is indicative of susceptibility to celiac disease and does not imply the development of the disease, the diagnosis of which must be verified with serological blood tests and a specific intestinal biopsy.

**Absence of HLA at risk for celiac disease**  
**This condition makes the onset of celiac disease highly unlikely**

For any further clarification regarding the interpretation of the results, contact an accredited facility or the Reference Center for the diagnosis of celiac disease.

Scientific Director

## DESCRIPTION OF CELIAC DISEASE

The term celiac disease derives from Coeliacus, in turn derived from the Greek Koiliakós which means «those who suffer in the Intestines» from Koilia (i.e. «abdomen, belly»). A term introduced in the 1st century. A.D. by a famous doctor of the time, Aretaeus of Cappadocia. Celiac disease is a GENETIC AUTOIMMUNE DISEASE of the small intestine of multi-faceted implications.

## CHARACTERISTICS

- Permanent hypersensitivity to gluten linked to an alteration of the immune system. Specifically, it is an immune-mediated enteropathy characterized by protein intolerance of gluten (gliadins) present in wheat, rye and barley.
- This Alteration can arise in different periods of life. The wide variability in the appearance of symptoms from one person to the next, does not allow adequate epidemiological studies.
- The only possible therapy is the total and permanent exclusion of cereals containing gluten from the diet.
- It is the most common form of "food disease " in the western world. In Europe and in the United States of America, the disease affects 0.5% to 1% of the population.
- The categories at risk are: family members of a celiac disease affected patient, patients suffering from non- autoimmune diseases not specifically related to any organ, and patients who present diagnostic difficulties.

## THE CAUSES

Celiac disease develops only in predisposed subjects following exposure to gluten; the gliadin contained in it, contains sequences of amino acids capable of sensitizing the lymphocytes of the layers in the intestine.

Three factors are needed to trigger celiac disease:

- Genetic predisposition.
- Presence of a good quantity of gluten in the diet.
- Intervention of triggering factors, such as viral infections or physical and psychological stress.

In multi-faceted diseases, no gene mutation in itself causes the disease, the genetic tests do not lead to a fullproof diagnosis but allow the identification of genetic characteristics which lead to an increased risk of developing a certain pathology (genetic susceptibility testing). From a genetic point of view, in celiac patients, variations (or alleles) can be found in some HLA system genes that alone, constitute the main genetic risk factors, in at least 40% of the increase of the prevalence of the disease in siblings (approximately 10% compared to 1% of the general population). There is also an increase in prevalence among parents and children of patients compared to the population in general, however to a lesser extent, as can be seen from some studies in which it has been quantified separately from that of the siblings.

### TYPICAL SYMPTOMS OF CELIAC DISEASE

Abdominal cramps
Bloating and abdominal distention
Colitis
Flatulence
Intermittent or chronic diarrhea

### ATYPICAL SYMPTOMS OF CELIAC DISEASE

Celiac disease tends to occur quite heterogeneously, meaning between an individual celiac and another there may be profound differences in symptoms related to the disease. In fact, the typical forms represent only the tip of the iceberg, the part of which is submerged (which represents the majority of cases) is made up of atypical or silent forms (they are considered silent forms with absent symptoms, despite the presence of typical lesions of the intestinal mucosa). To be precise, rather than atypical symptoms it would be more correct to talk about atypical signs of celiac disease and, even better, of potentially associated pathologies (and related complications).

### ATYPICAL SYMPTOMS AT THE INTESTINAL LEVEL

Abdominal discomfort
Flatulence
Impaired defecation
With bloating

### ATYPICAL ORAL SYMPTOMS

Atrophic glossitis
Hypoplasia of tooth enamel
Intraoral manifestations of dermatitis herpetiformis
Recurrent aphthous stomatitis

### ATYPICAL SYMPTOMS SPECIFIC TO FEMALE SEX

Alterations in the menstrual cycle
Amenorrhea
Early menopause
Endometriosis
Difficulty in conception
Late menarche
Recurrent miscarriages

### ATYPICAL SYMPTOMS SPECIFIC TO MALE SEX

Decrease in libido
Hypogonadism
Impotence
Oligospermia

### OTHER ATYPICAL SYMPTOMS OF CELIAC DISEASE

Alopecia areata	Constantly elevated transaminase values	Peripheral polyneuropathy
Anxiety, depression	Dementia	Psoriasis
Arthritis, arthralgia (joint pain)	Epilepsy	Pubertal delay
Asthenia	Follicular keratosis	Schizophrenia
Asthma	Hepatic steatosis	Spinocerebellar syndrome
Bronchial hyperactivity	Hypo/hyperthyroidism	Vitiligo
Cerebellar ataxia	Hyposplenism (thrombocytosis)	
Cognitive disorders	Osteopenia, osteoporosis	

Due to the various atypical manifestations described, celiac disease has earned the nickname "chameleon" disease.

### TREATMENT

The only possible treatment for celiac disease is a very strict and permanent gluten-free diet, which allows you to eliminate symptoms and reconstitute intestinal tissues, usually within 6-18 months from diagnosis. Following a gluten-free diet means changing your lifestyle and eliminating all flour-based foods made from wheat and barley (therefore: pasta and bread, pizza, rusks, breakfast cereals). Meat, vegetables, fruit, potatoes, rice and corn, legumes, do not contain gluten and therefore can enter the celiac diet easily. There are many gluten-free substitute foods on the market, which are specially formulated for celiacs and people with gluten intolerance. These products methods - replace wheat with a naturally gluten-free cereal or are deglutinated with chemical and/or physical - are tolerated by celiacs. The products bearing this claim and which are included in the National Register of foods from the Ministry of Health, can be purchased through the free supply quota which the National Health System recognizes for every celiac with the corresponding due amount which varies depending on sex and age. Furthermore, it must be considered that gluten can be "hidden" in foods, where it is added as an additive. If one of these foods is produced by avoiding adding gluten as an additive and/or taking care not to contaminate the packaged product with gluten during the production chain, it will lead to having a label with the nutritional indication "gluten-free - suitable for celiacs".