



*Genetic test*

# GLUTEN INTOLERANCE



**MICROGENOMICS**  
*Advanced Genomic Solutions*





## WHAT IS THE CELIAC DISEASE

Celiac disease is a food intolerance caused by hypersensitivity to gluten, a protein found in wheat and many other grains, such as rye, barley, spelled, oats, kamut.

In patients with celiac disease the ingestion of food containing gluten causes a reaction of the immune system, which attacks the small intestine cells while interfering with the absorption of nutrients found in these foods.

“ **Celiac disease is a chronic inflammation of the small intestine in GENETICALLY predisposed individuals.** ”

The main symptoms of people suffering from gluten intolerance include diarrhea, bloating and abdominal pain, vomiting, constipation and weight loss due to malabsorption of food.

Undiagnosed and untreated gluten hypersensitivity can also lead to serious diseases. During infancy, the most important period for growth and development of the child, it can cause problems such as difficulty of development and growth retardation; in adulthood you may have symptoms such as anemia, osteoporosis, depression or anxiety, infertility, miscarriages, absence of menstruation, but also more serious conditions such as dermatitis herpetiformis, diabetes and intestinal lymphoma.

An early diagnosis is crucial to start a timely therapy based on a completely gluten-free diet, thus improving the intestinal injury and avoiding the onset of serious complications.

“ **In Italy one in 100 people suffers from celiac disease, but it is estimated that about 73% of people suffering from celiac disease are not yet aware of it.** ”

## GLUTEN INTOLERANCE IS WRITTEN IN THE GENES

To this date the preferred diagnostic test for the identification of celiac disease is histological analysis, an invasive test that is carried out by intestinal biopsy.

Genetic testing however is non-invasive procedure and is performed on the buccal mucosa cells, obtained by rubbing a cotton swab inside the mouth.

The analysis consists in HLA typing and it is a genetic susceptibility test that evaluates the greater or lesser predisposition of an individual to develop celiac disease based on the presence/absence of risk factors (such as DQ2, DQ8).

The presence of the alleles DQ2 and/or DQ8 determines an increased risk for celiac disease, according to the different combinations, up to about 14 times that of the general population, while the absence of risk alleles makes the development of the disease quite unlikely.

The HLA genes are immutable throughout life.

Their typing can distinguish between presence or absence of genetic susceptibility to the disease long before the appearance of any symptoms. Therefore, analysis of HLA genes have mainly negative predictive value, as the absence of risk alleles means that the development of the disease is highly unlikely.

In case of positive outcome, however, it is strongly recommended the execution of an invasive (intestinal biopsy test) because HLA molecules alone are not sufficient to cause the disease, in fact it only appears upon exposure to environmental trigger factors and in presence of other genetic factors.

# WHEN IS TESTING RECOMMENDED

Genetic testing is recommended in case of:

- gastrointestinal disorders such as diarrhea, bloating and abdominal pain or constipation;
- anemia, osteoporosis, depression, anxiety and dermatitis herpetiformis;
- unjustified reduction in weight;
- diagnosed in childhood, avoiding initially invasive tests such as the intestinal biopsy;
- doubtful cases in adulthood;
- relatives with celiac disease, ability to identify cases of family members of 1st degree at risk (parents, children and siblings);
- patients with diseases associated with celiac disease (diabetes mellitus type 1, selective IgA deficiency, autoimmune thyroiditis, autoimmune hepatitis, Down's syndrome, Turner syndrome, Williams syndrome).

# MICROGENOMICS GENETIC TESTING LABORATORY

Genetic testing for gluten intolerance is a painless and easy test to perform and is recommended that it should be provided by Medical Genetics laboratories that meet SIGU (Italian Society of Human Genetics) accreditation requirements.

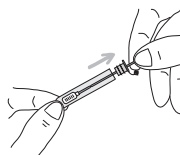
The Laboratory of Genetic analysis Microgenomics, UNI EN ISO 9001: 2008 certified and accredited to SIGUCert standards, will provide the test results in 10 working days identifying the presence or absence of increased risk for celiac disease.

You can take your saliva sample fast and pain free using the kit provided by our Laboratory.

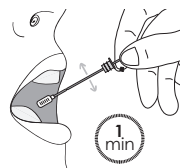
# SAMPLING KIT



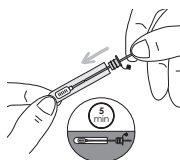
**1** Open the package and remove the pickup kit.



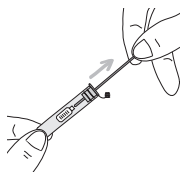
**2** Remove the swab from the tube with the cap. **Be careful not to touch the white pad.**



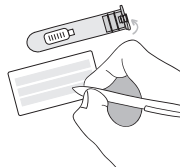
**3** Insert swab in the mouth and rub firmly against the inside wall of the cheek for at least 1 minute. Do the same with the second pad on the opposite cheek.



**4** Insert the swab into the tube and, without closing, let it dry for 5 minutes on a flat surface. **Never place the white pad on other surfaces.**



**5** Push the stopper inside the tube and, holding the latter firmly, pull the stick out to release the final white pad.



**6** Finally, seal the tube pushing the small cap. Apply the label with name, surname and date of birth on the tube.



**7** Complete and sign the consent form for the execution of the genetic test in its entirety.



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Microgenomics genetic testing laboratory is chartered  
to the Lombardy Region Health Service and certified  
ISO 9001:2008 e SIGUCert - Italian Society of Human Genetics.