Genetic test

LACTOSE INTOLERANCE

MICROGENOMICS

Advanced Genomic Solutions

WHAT IS LACTOSE INTOLERANCE

Lactose intolerance occurs when there is no partial or total lactase, an enzyme found in the small intestine. The function of lactase is to break down lactose, the main sugar in milk, into its two simple sugars: galactose and glucose. The first is essential for the formation of the nervous structures in the child, the second represents the body's primary energy substrate.
In case of shortage or total lack of this enzyme, the lactose is not digested and remains in the large intestine where it is fermented by the intestinal bacterial flora resulting in production of gas, water and acids.

The main symptoms of those suffering from lactose intolerance include diarrhea, nausea, bloating and abdominal pain, flatulence, tympanites, slow digestion, fatigue and rashes. These symptoms occur from 30 minutes to 2 hours after ingestion of foods containing lactose, but can be different from subject to subject, since their severity depends on each individual's level of lactase production shortage.

Lactose is present in different types of milk (cow, donkey and goat) in addition to breast milk, and all the products resulting from the processing of milk, like cheese, butter and cream. What not everybody knows is that it is often added to premade and commercial foods, as well as the most unexpected edibles, such as meat, sweets and frozen products.

> *"More than half the world's population is lactose intolerant and about 50% of the Italian population suffers from it."*

LACTOSE INTOLERANCE IS WRITTEN IN THE GENES

In 90% of cases, lactose intolerance is due, in Caucasians, to a variation of DNA: a polymorphism C/T - in position 13910, in the regulatory region of the lactase gene. If such a polymorphism is present in homozygous, ie in both copies of the gene, it can lead to a reduced expression of this enzyme in microvilli of the small intestine, causing lactase deficiency. This reduced expression means that the amount of undigested lactose is always more with the passing years.
Genetic testing for lactose intolerance thus enables to distinguish between those who have both normal
copies of the gene (T/T), some that have only one healthy (T/C) and those that have both mutated (C/C). TT homozygotes produce high levels of lactase and therefore are able to digest lactose without any problems; The C variant in homozygous (genotype C/C) is associated with a lower production of lactase causing a greater difficulty in digesting lactose.

The frequency of CC homozygotes in the North-European population is around 15-20% while it is around 50% in some Mediterranean populations (Italian, Greek, Spanish, etc.).
The genetic test then indicates whether or not the subject is predisposed to develop a deficiency of lactase (hypolactasia) later in life. So genetic testing is aimed at the identification of -13910 C > T variant in the lactase gene and therefore should be considered an exclusion test, that is useful to exclude the genetic component's involvement in the onset of any disturbances resulting from the ingestion of lactose-containing foods.

WHEN IS TESTING RECOMMENDED

Genetic testing is recommended in case of:

- Positive Breath test (breath test); in this case the genetic test can be useful to clarify whether it is a final condition (as what's "written" in DNA does not change in the course of life) or if it is a secondary, and then temporary hypolactasia;
- Presence of symptoms (gastrointestinal disorders such as diarrhea, nausea, bloating and abdominal pain, flatulence, bloating, slow digestion);
- Family history of lactose intolerance;
- Positive test of intolerance to dairy.

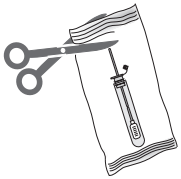
The test is also recommended as a form of prevention in newborns, as an alternative to more complex Breath tests.

MICROGENOMICS GENETIC TESTING LABORATORY

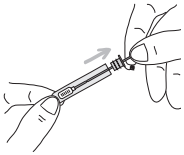
Microgenomics Laboratory's genetic testing for lactose intolerance is very accurate, rapid, non-invasive, painless and easy to perform. For its execution it is not necessary to ingest lactose (scheduled for the practice of Breath testing) and, in contrast to the conventional functional tests, fasting is not necessary.
By using the kit provided by our Laboratory you can take your saliva sample, by rubbing the cotton swab inside your mouth, fast and pain free.

In 10 days Microgenomics Lab will provide you the results that identify the presence or absence of -13910 C>T variant.

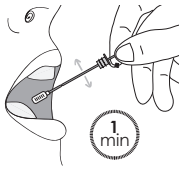
SAMPLING KIT



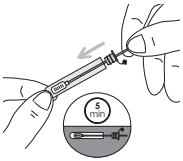
1 Open the package and remove the pickup kit.



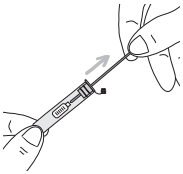
2 Remove the swab from the tube with the cap. **Be careful not to touch the white pad.**



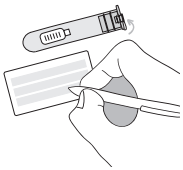
3 Insert swab in the mouth and rub firmly against the inside wall of the cheek for at least 1 minute. Do the same with the second pad on the opposite cheek.



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.

MICRO GENOMICS
ADVANCED GENOMIC SOLUTIONS

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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.