synlab Budapest Diagnostic Center
Genoid Molecular Diagnostic Laboratory

19. Szanatórium u.
H-1121 Budapest, Hungary

Lab: +36 1 465 01 24

www.synlab.hu
www.genoid.hu

Lactose intolerance

genetic testing

HU-20-00-4_84_Laktoseintoleranz

What does lactose intolerance mean?

Lactose, a sugar found in milk, is degraded in the intestines by an enzyme called lactase. This process is vital for infants, while lactase activity normally decreases after weaning. There are people, however, whose genetic makeup permits sustained lactase activity, thus the ability to digest milk. For conventional reasons, they are today regarded as "normal", whereas people who are not able to digest lactose in their adulthood are termed lactose intolerant.

The frequency of adult type lactose intolerance is 5 to 70% in Europe and 80% to 100% in Asia and Africa.

What are the types of lactose intolerance?

- The abovementioned "adult type lactose intolerance" is called primary because it is the consequence of a downregulation of lactase activity in the intestinal mucosa, which is associated in 93% of the cases with the C/T-13910 polymorphism in the regulatory region of the lactase gene.*
- Secondary or acquired lactose intolerance is usually related to intestinal mucosal damage caused by an underlying disease or state, is accompanied by other malabsorption symptoms and is often transient.
- Other milk-related disorders include the very rare congenital lactase deficiency, in which a genetic error results in the total absence of functional lactase enzyme causing severe symptoms as early as at the first breastfeeding. Milk protein allergy should also be distinguished from lactose intolerance.

What are the symptoms of adult type lactose intolerance?

The most common symptoms are abdominal pain, diarrhoea, bloating, and nausea after consumption of milk or dairy products. The disorder usually manifests in children older than 5 years of age, occasionally only around 20 years of age.

How can it be diagnosed?

The diagnosis is traditionally based on the measurement of the quantity of exhaled hydrogen after lactose load. Lactose consumption, however, can be very unpleasant, and also, the test is not sufficiently accurate and should be repeated from time to time. Fortunately research in the past decade lead to better understanding of the underlying genetics, which makes it possible to diagnose adult type lactose intolerance by a discomfort-free, more accurate and rapid genetic testing method.*

Genetic testing of lactose intolerance:

- **no discomfort**
- **sound result**
- **valid for lifetime**

This test has to be performed only once, because it gives definite information on the state of lactose tolerance or intolerance.

For whom is lactose intolerance genetic testing useful?

For those older than 5 years of age with gastrointestinal symptoms after dairy consumption; for those unsure about a connection between milk consumption and their discomfort; for those who simply want to know what they have inherited from their parents. Children under 5 years of age with frequent abdominal complaints (cramps, diarrhoea), the test can be performed to rule out lactose intolerance.

What is the procedure of testing?

All that is necessary is a cheek swab that can be obtained easily even by oneself. Blood sample in a purple-top (EDTA) tube is also suitable. The result is complete within 10 work days after receipt of sample and the report includes interpretation in a plain language.

** Rasinperä et al, Gut 2004;53:1571*