DNK s.r.o.
diagnostické centrum

Laboratory diagnosis of genetic predisposition to lactose intolerance

Diagnostic kit to detect polymorphism in the MCM6 gene using real-time PCR

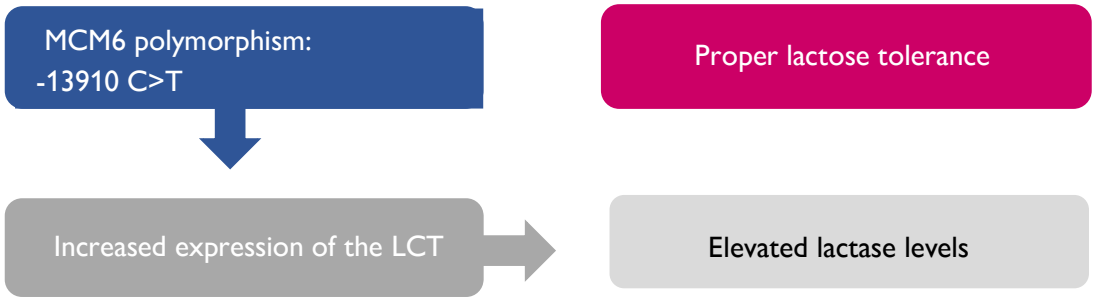
Lactose (milk sugar) is a carbohydrate from the group of disaccharides found in mammalian milk. It promotes the absorption of calcium and other minerals in the intestines, as well as the reproduction of normal intestinal flora.

Lactase is an enzyme that promotes the absorption of lactose in the gastrointestinal tract (GIT). This enzyme reaches its maximum activity at the time of birth of the child, after 2-2.5 years of life, lactase activity in carriers of the normal genotype (C/C) gradually decreases. This is called primary or age-related lactose intolerance due to hereditary lactase deficiency.

Lactase deficiency (lactose intolerance) is the inability of the body to breakdown lactose, accompanied by digestive disorders. Often this condition leads to the development of calcium deficiency, which increases the risk of osteoporosis. Lactose intolerance is generally the population norm and occurs in 3/4 of the adult world population. In a number of populations, due to the long-term use of dairy products as an important part of the diet, genetic mutations have spread, which make it possible not to turn off lactase expression throughout life.

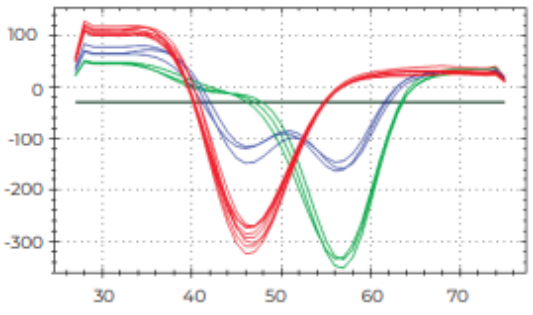
The most common mutation is single-nucleotide polymorphism (SNP) of the MCM6 gene: -13910 C>T, which is associated with a change in lactase activity and is being studied to identify a genetic predisposition to lactase deficiency.

Clinical significance of MSM6 SNP detection: -13910 C/T



Diagnostic kit RealBest-Genetics MCM6 (Cat. No. 3804)

- Determination of SNP -13910 C/T in *MCM6* gene
- The principle of SNP detection is based on amplification of a selected fragment of human DNA and subsequent detection of melting curves of hybrid complexes of PCR products and specific probes with fluorescent tag
- The diagnostic kit is designed for 48 samples, including control samples and for use with Real-Time devices CFX96 (BioRad, USA), Gentier 96E/R (Xi'an TianLong, Science and Technology Co., Ltd., China).



Indications for molecular-genetic testing

• Symptoms of lactose intolerance (indigestion after consuming milk and dairy products) in order to choose a diet

• In the differential diagnosis of lactose intolerance and other diseases of the digestive tract

• Assessment of the risk of developing osteoporosis, especially in menopausal women

• Assessment of the likelihood of lactose intolerance in young children (for the prevention of bowel disorders)

Features and advantages of the diagnostic kit

- Ready Master Mix for PCR: lyophilized, simplification of the PCR analysis procedure and high stability of test quality
- Specimens: whole blood or buccal epithelium
- High stability of the kit: storage at a temperature of 2–8 ° C; transport up to 26 °C for not more than 10 days

Interpretation of analysis results

Genotype	Possible manifestations of SNP	Additional risk factors
C/C	Increases the risk of lactose intolerance and osteoporosis	1. Lack of calcium in food 2. Postmenopause (women) 3. Age 4. Intestinal infections, celiac disease, Crohn's disease
C/T	It is associated with variable levels of lactose activity and increases the risk of secondary lactose intolerance	
T/T	Increases the likelihood of proper lactose tolerance	-

Diagnostické centrum DNK, s.r.o.
Brestová 14, 821 02 Bratislava
+421 911 299 324, +421 911 211 404
dnk@pharma.sk, diagnostika@pharma.sk
www.pcr.sk