

LCT POLYMORPHISMS LACTOSE INTOLERANCE

ORDERING INFORMATION

REF: GEN-024-25 RDM Code: 2256381/R
Tests: 25 Reactions: 31 x 2
REF: GEN-024-50 RDM Code: 2145488/R
Tests: 50 Reactions: 62 x 2
CND Code: W0106010499
Manufacturer: BioMol Laboratories s.r.l.

CONTENTS OF THE KIT

The kit consists of reagents for
Real-Time PCR amplification
*reagents for the extraction of
genomic DNA are not supplied
in the kit

For in vitro diagnostic use



PRODUCT CHARACTERISTICS

Device belonging to the family of in vitro medical devices **REAL-TIME QUALITATIVE PCR-GENETIC VARIANTS**. Detection of -13910 C>T and -22018 G>A polymorphisms of the gene encoding the enzyme lactose-phenyloxy hydrolase (LPH) by Real-Time PCR technique. Optimized kit for Real-Time PCR instrumentation Biorad CFX96 Dx, Biorad Opus Dx, Agilent AriaDx, Hyris bCUBE and Hyris bCUBE3 with Hyris bAPP.

SCIENTIFIC BACKGROUND

Lactose is the main sugar in milk, and lactose intolerance (LI) is very common. Symptoms of LI include diarrhea, abdominal pain, and flatulence after drinking or eating milk or products containing milk. These symptoms are caused by low levels of intestinal lactase due to mucosal injury or, more often, due to reduced genetic expression of the enzyme lactose-phenyloxy hydrolase (LPH).

CLINICAL SIGNIFICANCE

Lactose intolerance (LI) is inherited as an autosomal recessive trait that causes reduction in the enzymatic activity of lactose-phenyloxy hydrolase (LPH) in intestinal cells, causing a decrease in the ability to convert lactose into the absorbable sugars glucose and galactose. Decline in LPH enzyme activity is known to occur at age 12, however a portion of individuals retain neonatal LPH activity by exhibiting lifelong lactose tolerance (LT). Lactase persistence varies among different human populations, from 95% in Northern Europeans and North Americans to about 50% or less in South American and African countries, such as Cameroon, Mali, and South Africa, to about 0% in some Asian countries, including China. The literature reports that in addition to biochemical blood analyses, genetic markers can be useful for the diagnosis of LI. To date, two main markers have been identified: single nucleotide polymorphisms (SNPs) C>T-13910 (rs4988235) and G>A-22018 (rs182549), located upstream of the lactase gene (LCT). In heterozygous -13910 C/T individuals, the 50% reduction in lactase activity level is normally sufficient to ensure digestion of lactose. Individuals with the 13910T/T genotype are perfectly tolerant to lactose while if the -13910 mutation is present in the homozygous state (C/C) there is a total deficiency of the lactase enzyme in adults. 100% of individuals with primary lactose intolerance (IPL) have the -13910 C/C genotype. Approximately 90% of these also have the -22018 G/G genotype while the remaining 10% have the -22018 G/A or A/A genotype, generally with milder symptoms. Recent is the relationship between lactase activity and vitamin D and calcium levels.

§ Nutrients. 2024 Sep 5;16(17):3002. doi: 10.3390/nu16173002. Bone Mineral Density and the Risk of Type-2 Diabetes in Postmenopausal Women: rs4988235 Polymorphism Associated with Lactose Intolerance Effects

§ Int J Mol Sci. 2023 Jun 15;24(12):10191. doi: 10.3390/ijms241210191. A Comprehensive Look at the -13910 C>T LCT Gene Polymorphism as a Molecular Marker for Vitamin D and Calcium Levels in Young Adults in Central and Eastern Europe: A Preliminary Study

§ United European Gastroenterol J. 2019 Mar;7(2):210-216. doi: 10.1177/2050640618814136. Epub 2018 Nov 15. 13910C>T and 22018G>A LCT gene polymorphisms in diagnosing hypolactasia in children

§ Association of lactase 13910 C/T polymorphism with bone mineral density and fracture risk: a meta-analysis. J Genet. 2017. Dec;96(6):993-1003. doi: 10.1007/s12041-017-0866-8.

§ BMJ Open. 2011 Jul 29;1(1):e000125. doi: 10.1136/bmjopen-2011-000125.

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DESCRIPTION	LABEL	VOLUME		STORAGE
		GEN-024-25	GEN-024-50	
Mix oligonucleotides and probes	Mix LCT -13910 C>T 10X	1 x 77,5 µl	2 x 77,5 µl	-20°C
Mix oligonucleotides and probes	Mix LCT -22018 G>A 10X	1 x 77,5 µl	2 x 77,5 µl	-20°C
Mix buffer and Taq polymerase enzyme	Mix Real-Time PCR 2X	1 x 775 µl	2 x 775 µl	-20°C
Deionized H ₂ O	Deionized H ₂ O	1 x 1 ml	1 x 1 ml	-20°C
Genomic DNA or recombinant DNA Control 1	Control 1 CC/GG	1 x 35 µl	2 x 35 µl	-20°C
Genomic DNA or recombinant DNA Control 2	Control 2 CT/GA	1 x 35 µl	2 x 35 µl	-20°C
Genomic DNA or recombinant DNA Control 3	Control 3 TT/AA	1 x 35 µl	2 x 35 µl	-20°C

TECHNICAL CHARACTERISTICS

COD. GEN-024-25 / COD. GEN-024-50

STABILITY	18 months
REAGENTS STATUS	Ready to use
BIOLOGICAL MATRIX	Genomic DNA extracted from whole blood, tissue, cells
POSITIVE CONTROLS	Recombinant DNA for at least 3 analytical sessions (GEN-024-25) Recombinant DNA for at least 6 analytical sessions (GEN-024-50)
TECHNOLOGY	Real-time PCR; oligonucleotides and specific probes; 2 FAM/HEX fluorescence channels
VALIDATED INSTRUMENTS	Biorad CFX96 Dx, Biorad Opus Dx, Agilent AriaDx, Hyris bCUBE and Hyris bCUBE3 with Hyris bAPP.
RUNNING TIME	85 min
THERMAL CYCLING PROFILE	1 cycle at 95 °C (10 min); 45 cycles at 95 °C (15 sec) + 60 °C (60 sec)
ANALYTICAL SPECIFICITY	Absence of non-specific pairings of oligonucleotides and probes; absence of cross-reactivity
LIMIT OF DETECTION (LOD)	≥ 0,016 ng of genomic DNA
LIMIT OF BLANK (LOB)	0% NCN
REPRODUCIBILITY	99,9%
DIAGNOSTIC SPECIFICITY / DIAGNOSTIC SENSITIVITY	100%/98%