# LCT POLYMORPHISMS
# LACTOSE INTOLERANCE

For in vitro diagnostic use

CE IVD

## ORDERING INFORMATIONS

*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*

## PRODUCT CHARACTERISTICS

Detection of -13910 C>T and -22018 G>A polymorphisms of the gene encoding the enzyme lactose-phenytoin 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 milk-containing products. These symptoms are caused by low intestinal lactase levels due to mucosal lesions or, more often, due to reduced genetic expression of the enzyme lactose-phenytoin hydrolase (LPH).
Lactose intolerance (LI) is inherited as an autosomal recessive trait that results in decreased enzymatic activity of lactose-phenytoin hydrolase (LPH) in intestinal cells, resulting in decreased ability to convert lactose into absorbable sugars glucose and galactose.

*§ Frequency of LCT-13910C/T and LCT-22018G/A single nucleotide polymorphisms associated with adult-type hypolactasia/lactase persistence among Israelis of different ethnic groups. Gene. 2013 Apr 25; 519 (1):67-70. doi: 10.1016/j.gene.2013.01.049. Epub 2013 Feb 13.*

*§ The European lactase persistence genotype determines the lactase persistence state and correlates with gastrointestinal symptoms in the Hispanic and Amerindian Chilean population: a case-control and population-based study.*

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

*§ LCT-13910C>T polymorphism-associated lactose malabsorption and risk for colorectal cancer in Italy. Dig Liver Dis. 2010 Oct; 42 (10):741-3. doi: 10.1016/j.dld.2010.02.013. Epub 2010 Apr 2.*

*§ 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.*

## CLINICAL SIGNIFICANCE

LPH enzyme activity decline is known to occur by age 12, however a proportion of individuals retain neonatal LPH activity by showing lifelong lactose tolerance (LT). Lactase persistence varies in human populations, ranging 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 analyzes of blood, genetic markers can be useful for the diagnosis of LI. To date, two major markers have been identified: the single nucleotide polymorphisms (SNPs) C>T-13910 (rs4988235) and G>A-22018 (rs182549), located upstream of the lactase gene (LCT). In individuals heterozygous -13910 C/T, a 50% reduction in lactase activity level is usually 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 the adult. 100% of individuals with primary lactose intolerance (IPL) have the -13910 C/C genotype. About 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.

BIOMOL LABORATORIES S.R.L.
Via Arcora 110 (Palazzo Gecos)
80013 Casalnuovo di Napoli, NA
info@biomollaboratories.com
biomollaboratories.it

ISO 9001:2015
ISO 13485:2016



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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 85 µl | 1 x 170 µl | -20°C |
| Mix oligonucleotides and probes | Mix LCT -22018 G>A 10X | 1 x 85 µl | 1 x 170 µl | -20°C |
| Mix buffer and Taq polymerase enzyme | Mix Real-Time PCR 2X | 1 x 850 µl | 2 x 850 µl | -20°C |
| Deionized $H_20$ | Deionized $H_20$ | 2 x 1 ml | 2 x 1 ml | -20°C |
| Genomic DNA or recombinant DNA | Control + 1 | 1 x 45 µl | 1 x 45 µl | -20°C |
| Genomic DNA or recombinant DNA | Control + 2 | 1 x 45 µl | 1 x 45 µl | -20°C |
| Genomic DNA or recombinant DNA | Control + 3 | 1 x 45 µl | 1 x 45 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 CONTROL | Recombinant DNA for at least 3 analytical sessions |
| 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 |
| ANALYTICAL SENSITIVITY : LIMIT OF DETECTION (LOD) | ≥ 0,016 ng of DNA |
| ANALYTICAL SENSITIVITY : LIMIT OF BLANK (LOB) | 0% NCN |
| REPRODUCIBILITY | 99,9% |
| DIAGNOSTIC SPECIFICITY / DIAGNOSTIC SENSITIVITY | 100%/98% |

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