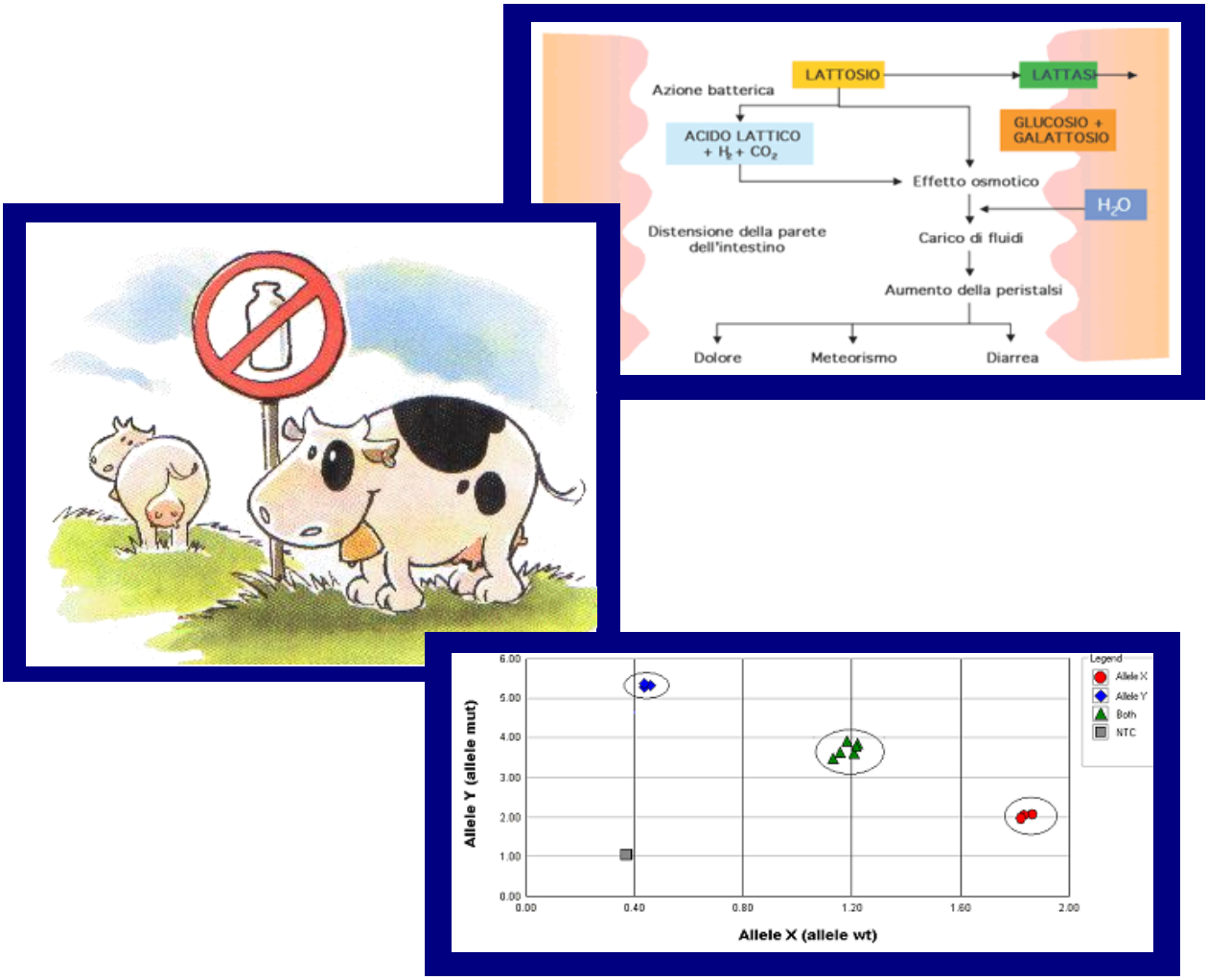
System for analysis of C13910T polymorphism in LCT gene associated to lactose intolerance by Real Time PCR.

LCT MUTATION KIT - RQ

cod. LC.01RQ



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Lactose is digested in the small intestine by the enzyme **lactase** (phlorizin hydrolase), a beta-galactosidase present on brush border of enterocytes lining the apex of the villi; lactase hydrolyzes lactose into D-glucose and D-galactose and compare as early as the 23rd week of gestation. Its activity increases during pregnancy reaching the maximum at birth; it remains maxim for the entire period in which breastfeeding is the exclusive nourishment of the child and, starting from weaning, begins to decrease according to a genetically defined scheme.

We talk about lactose intolerance when the subject is unable, or it is partially able, to digest the lactose taken from food.

Lactose intolerance or **adult-type hypolactasia** affects more than half of world's population. In Italy, the population which suffers of lactase deficiency varies between 15 and 40%; the frequency increases going from North to South of the peninsula and is particularly high in the Naples area.

The **symptoms** are substantially of gastrointestinal nature, but the symptomatology is not specific since it is manifested as well in the presence of intolerance of other nature and also with food allergies.

Recently, has been identified a single nucleotide polymorphism (**SNPs**) located at 13910 (**C-13910T**) upstream the lactase gene (**LCT**), in intron 9 of the adjacent gene MCM6 (MiniChromosome Maintenance 6), an enhancer of the promoter of LCT gene.

The 13910T allele of this polymorphism showed a strong association with lactase persistence in European populations; the ***CT*** and **TT genotypes** are associated with lactase **persistence** while the ***CC* genotype** is associated to **hypolactasia** of the adult.

Genotyping of this polymorphism allows to make a differential diagnosis of the genetic causes, against the secondary ones, of lactose intolerance.

How does the kit function?

The **LCT mutation RQ-kit** allows the discrimination of **C-13910T** polymorphism in LCT gene associated to lactose intolerance from DNA extracted from peripheral blood or buccal swab by Real Time PCR.

The kit contains, in addition to primers that allow the amplification of the gene region containing the polymorphism and probes that allow the detection of the two alleles, also the Master Mix for the amplification and positive controls.

SNP: rs 4988235

Starting samples: peripheral blood, buccal swabs

DNA extraction method: QIAamp DNA blood mini kit, QIAcube, QIAsymphony (Qiagen), High Pure PCR template preparation kit (Roche), Nuclisens EasyMAg (Biomerieux)

Real Time PCR Systems: Real Time PCR System series 7000, Step one/plus (Applied Biosystems), Rotor-Gene 3000, 6000, Q (Qiagen), CFX96 (BioRad), SmartCycler System (Cepheid)

Fluorophores: 6-FAM, VIC

Kit content

Label	Content
LCT-RQ Master Mix	Mix for amplification of LTC gene
LCT1 Pr T-FAM	Probe complementary to T allele
LCT1 Pr C -VIC	Probe complementary to C allele

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