

## SNP SUMMARY: LACTASE (LCT) -13910 C>T (or MCM6)

Overview	Lactose intolerance or lactase non-persistence (LNP) is the syndrome of diarrhea, abdominal pain, or flatulence, occurring after lactose ingestion. These symptoms are caused by a decreased ability to hydrolyze lactose due to a decrease in lactase expression, which is associated with a gene variation upstream from the lactase gene.		
Name of gene	Lactase gene		
Symbol of gene	LCT		
Gene database	http://www.ncbi.nlm.nih.gov/SNP/snp_ref.cgi?rs=rs4988235		
rs number	rs4988235		
Base change	C>T		
Position on gene	-13910 upstream of the lactase transcriptional start site. The SNP is located in an intron of the adjacent gene minichromosome maintenance 6 (MCM6).		
SNP's nomenclature	<i>LCT</i> -13910 C>T		

<ul> <li>hydrolyze lactose.</li> <li>Leading to a condition known as Lactose intolerance/non-persistence.</li> <li>Not all mechanisms as to how gene expression is turned off or reduced are known but possible explanations involve th activator or repressor molecules.</li> <li>Various studies have demonstrated that the −13910 C&gt;T SNP is functional and is associated with changes in LCT gen expression.</li> </ul>						
African/African American CC=79.2%, CT=20.8%, TT=0%     Hispanic CC=52.2%, CT=39.1%, TT=8.7%    Hispanic CC=52.2%, CT=39.1%, TT=8.7%    Lactase or lactase-phlorizin hydrolase (LPH) enzyme    Lactase or lactase-phlorizin hydrolase (LPH) enzyme    Lactase gene expression is turned off after the weaning phase and therefore individuals may have a decreased ability thydrolyze lactose.   Leading to a condition known as Lactose intolerance/non-persistence.   Not all mechanisms as to how gene expression is turned off or reduced are known but possible explanations involve the activator or repressor molecules.   Various studies have demonstrated that the -13910 C>T SNP is functional and is associated with changes in LCT gene expression.   Individuals homozygous for the C allele (CC genotype) have almost undetectable levels of intestinal lactase production compared to TC or TT individuals.    Mittp://interactive-biology.com/wp-content/uploads/2012/08/Lactase-hydrolysis1-1024x232.png   CH <sub>2</sub> OH	Population frequency	It is found predominantly in European / Caucasian populations				
Name of enzyme  Lactase or lactase-phlorizin hydrolase (LPH) enzyme  Lactase or lactase-phlorizin hydrolase (LPH) enzyme  Lactase gene expression is turned off after the weaning phase and therefore individuals may have a decreased ability thydrolyze lactose.  Leading to a condition known as Lactose intolerance/non-persistence.  Not all mechanisms as to how gene expression is turned off or reduced are known but possible explanations involve th activator or repressor molecules.  Various studies have demonstrated that the -13910 C>T SNP is functional and is associated with changes in LCT genexpression.  Individuals homozygous for the C allele (CC genotype) have almost undetectable levels of intestinal lactase production compared to TC or TT individuals.  Biochemical pathway  http://interactive-biology.com/wp-content/uploads/2012/08/Lactase-hydrolysis1-1024x232.png  CH2OH  Lactase  Lactase in hydrolysed into D-galactose and D-glucose in the presence of adequate lactase enzyme.		• Caucasian CC=26.7%, CT=13.3%, TT=60%				
Lactase or lactase-phlorizin hydrolase (LPH) enzyme		African/African American CC=79.2%, CT=20.8%, TT=0%				
Impact of SNP on biological pathway  - Lactose is hydrolysed by the enzyme lactase into monosaccharides galactose and glucose utilised for energy.  - Lactase gene expression is turned off after the weaning phase and therefore individuals may have a decreased ability thydrolyze lactose.  - Leading to a condition known as Lactose intolerance/non-persistence.  - Not all mechanisms as to how gene expression is turned off or reduced are known but possible explanations involve the activator or repressor molecules.  - Various studies have demonstrated that the -13910 C>T SNP is functional and is associated with changes in LCT genexpression.  - Individuals homozygous for the C allele (CC genotype) have almost undetectable levels of intestinal lactase production compared to TC or TT individuals.  Biochemical pathway  http://interactive-biology.com/wp-content/uploads/2012/08/Lactase-hydrolysis1-1024x232.png  - CH2OH OH O		Hispanic CC=52.2%, CT=39.1%, TT=8.7%				
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Clactase gene expression is turned off after the weaning phase and therefore individuals may have a decreased ability to hydrolyze lactose.  Leading to a condition known as Lactose intolerance/non-persistence.  Not all mechanisms as to how gene expression is turned off or reduced are known but possible explanations involve the activator or repressor molecules.  Various studies have demonstrated that the -13910 C>T SNP is functional and is associated with changes in LCT genexpression.  Individuals homozygous for the C allele (CC genotype) have almost undetectable levels of intestinal lactase production compared to TC or TT individuals.  Biochemical pathway  http://interactive-biology.com/wp-content/uploads/2012/08/Lactase-hydrolysis1-1024x232.png  CH2OH  OH  OH  OH  OH  OH  OH  OH  OH  OH	-	Lactose is hydrolysed by the enzyme lactase into monosaccharides galactose and glucose utilised for energy.				
Not all mechanisms as to how gene expression is turned off or reduced are known but possible explanations involve th activator or repressor molecules.      Various studies have demonstrated that the -13910 C>T SNP is functional and is associated with changes in LCT genexpression.      Individuals homozygous for the C allele (CC genotype) have almost undetectable levels of intestinal lactase production compared to TC or TT individuals.    http://interactive-biology.com/wp-content/uploads/2012/08/Lactase-hydrolysis1-1024x232.png	biological pathway	<ul> <li>Lactase gene expression is turned off after the weaning phase and therefore individuals may have a decreased ability to hydrolyze lactose.</li> </ul>				
activator or repressor molecules.  • Various studies have demonstrated that the -13910 C>T SNP is functional and is associated with changes in LCT gen expression.  • Individuals homozygous for the C allele (CC genotype) have almost undetectable levels of intestinal lactase production compared to TC or TT individuals.  Biochemical pathway  http://interactive-biology.com/wp-content/uploads/2012/08/Lactase-hydrolysis1-1024x232.png  CH2OH  OH  OH  OH  OH  OH  OH  OH  OH  OH		Leading to a condition known as Lactose intolerance/non-persistence.				
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Biochemical pathway  http://interactive-biology.com/wp-content/uploads/2012/08/Lactase-hydrolysis1-1024x232.png  CH2OH OH OH Lactase H2OH OH OH Lactase Lactose is hydrolysed into D-galactose and D-glucose in the presence of adequate lactase enzyme.		<ul> <li>Various studies have demonstrated that the −13910 C&gt;T SNP is functional and is associated with changes in LCT gene expression.</li> </ul>				
Nutrient interaction  CH2OH OH OH Lactose Lactose  Lactose is hydrolysed into D-galactose and D-glucose in the presence of adequate lactase enzyme.		individuals nemozygous for the c units (see genetype) have announced individuals levels of intestinal lactace production				
Nutrient interaction  Lactose is hydrolysed into D-galactose and D-glucose in the presence of adequate lactase enzyme.	Biochemical pathway	http://interactive-biology.com/wp-content/uploads/2012/08/Lactase-hydrolysis1-1024x232.png				
		CH <sub>2</sub> OH OH OH OH OH OH OH OH				
Established diet-gene Identification of a CC genotype is likely to predict the presence of lactase non-persistence (lactose intolerance), a TT genotype	Nutrient interaction	Lactose is hydrolysed into D-galactose and D-glucose in the presence of adequate lactase enzyme.				
	Established diet-gene	Identification of a CC genotype is likely to predict the presence of lactase non-persistence (lactose intolerance), a TT genotype				

interactions	will frequently, but not perfectly, predict lactase persistence, and a TC heterozygote genotype will not predict the phenotype, but should warrant further investigation.				
Potential dietary recommendations	<ul> <li>General tolerance level = 12g of lactose.</li> <li>Intake of cheese and yoghurt is better tolerated, large amounts of milk consumption is restricted.</li> <li>Lactase enzymes can be ingested concomitantly with lactose-containing food products.</li> </ul>				
Lactose content in various foods	Lactose containing food	Household quantity and grams (g) of lacotse	Quantity of food product providing approximately 12 g of lactose		
	Milk	250 ml/1 cup = 12 g	250 ml/1 cup		
	Yogurt	125 g (1 small tub) = 3.0-3.75 g	375 g		
	Cream	100 g = 3 g	400 g		
	Ice cream	125ml/1/2 cup = 6 g	250ml/1 cup		
	Cottage cheese, low fat	1/2 cup = 4-5 g	250ml/1 cup		
	Cheddar cheese	30g (matchbox-size) = 0 g	360 g		
	American, Swiss, or Parmesan cheese	30 g (matchbox-size) = 0-1 g	360 g		
Examples of lactose- containing food consumption	Providing approximately 12 g of lactose:  • 1 cup of milk and 30 g of Cheddar cheese OR  • ½ cup of Ice-cream and ½ cup of milk and 30g of cheddar cheese OR  • 1 tub 125g of yogurt and ½ cup of milk and ½ cup of low fat cottage cheese				



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