LACTASE PERSISTENCE AND LACTASE NON-PERSISTENCE
Prevalence, influence on body fat, body height
and relation to the metabolic syndrome

av

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Akademisk avhandling

Avhandling för medicine doktorsexamen i medicinsk vetenskap,
som enligt beslut av rektor kommer att föras offentligt
onsdag den 15 december 2010 kl. 09.00,
Wilandersalen, Örebro Universitetssjukhus

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Abstract

A single nucleotide polymorphism (SNP) in chromosome 2 gives humans the capability to continue digesting lactose after weaning. Among individuals of European descent it is the SNP (LCT-13910 C>T), which is located 14 kb upstream from the start of transcription of the lactase gene (LCT). A C to T mutation enables the continued production of the enzyme lactase throughout life, and thereby the digestion of lactose throughout life without symptoms of lactose intolerance. The trait is called lactase persistence (LP). LP is an autosomal dominant trait. In contrast, lactase non-persistent (LNP) individuals show a decline of lactase production after weaning. LNP individuals habitually show symptoms of lactose intolerance after consumption of milk and some milk products.

Using the LCT -13910 C>T SNP we reassessed the prevalence of LP/LNP in Sweden. We increased the accuracy of our estimates by using Hardy-Weinberg’s formula for allelic frequencies. We found a prevalence of 14% for LNP. This is about 5.5 fold higher than the prevalence formerly assumed for Sweden (range: 1-5%).

Childhood milk consumption has become normative since the beginnings of the last century. Studies using milk supplements in schools, performed in the UK in the 1920s, showed that childhood milk consumption led to gains in body weight and height in children. We readdressed this question given the changed socioeconomic settings a century later. In today’s, nutritionally replete socioeconomic settings childhood obesity is, instead of stunting and undernourishment, the target of public health nutrition. We did not find evidence for higher measures of body fat coupled to consumption of milk and milk products or the lactase genotype in Swedish children and adolescents. We found, nevertheless, a positive association between milk intake as well as LP and body height in Swedish children and adolescents.

Finally, we studied if LP/LNP, using Mendelian randomization (MR), affects the development of the metabolic syndrome in adults. We chose an accessible population from the Canary Islands in Spain with a prevalence of 40% for LNP and 60% for LP. We found a positive association of LP with metabolic syndrome. LP subjects of the Canary Islands exhibited a 57% higher risk to develop metabolic syndrome compared to LNP subjects. Interestingly, LP women showed a 93% higher risk to develop MS compared to a 22% higher risk in LP men.

In conclusion, the previously not known relatively high prevalence of LNP for Sweden impacts health care and public health policy decisions. Childhood milk consumption affects longitudinal growth, but not body fat mass in nutritionally replete countries like Sweden. The long-term effects of childhood milk consumption need to be elucidated. Non-caloric milk constituents might influence the somatotropic axis of children, which can have both positive and negative long-term effects. LP status and milk consumption may increase the susceptibility to develop metabolic syndrome in some populations. Non-caloric constituents of milk might exert sex specific effects increasing the risk to develop metabolic syndrome in susceptible populations.

Keywords: LCT -13910 C>T polymorphism, prevalence, lactase persistence and lactase non-persistence, milk and milk product intake, body fat, body height, metabolic syndrome

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