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Lactase persistence versus lactose intolerance: Is there an intermediate phenotype?

Dzialanski Z[1], Barany M[2], Engfeldt P[3], Magnuson A[4], Olsson LA[5], Nilsson TK[6].

Author information

1 University Health Care Research Center, School of Health and Medical Sciences, Örebro University, SE 701 82 Örebro, Sweden. Electronic address: zbigniew.dzialanski@regionorebrolan.se.

2 Department of Clinical Physiology, School of Health and Medicine Sciences, Örebro University, SE 701 82 Örebro, Sweden.

3 University Health Care Research Center, School of Health and Medical Sciences, Örebro University, SE 701 82 Örebro, Sweden.

4 Clinical Epidemiology and Biostatistics, School of Health and Medicine Sciences, Örebro University, SE 701 82 Örebro, Sweden.

5 Department of Clinical Chemistry, School of Health and Medical Sciences, Örebro University, SE 701 82 Örebro, Sweden.

6 Department of Medical Biosciences/Clinical Chemistry, Faculty of Medicine and Health, Umeå University, SE 901 87 Umeå, Sweden.

Abstract

BACKGROUND: According to the prevailing theory about the genetic background to lactose intolerance, there are three genotypes but only two adult physiological phenotypes: lactase persistence in individuals with the CT and TT genotypes and lactase non-persistence in individuals with the CC genotype. However, analysis of lactase activity from intestinal biopsies has revealed three distinct levels of activity, suggesting that an intermediate physiological phenotype may exist.

AIM: To assess possible disparities between different genotypes with regard to biomarkers of lactase activity and physical symptoms during an oral lactose load test.

METHODS: A retrospective study using an oral lactose load test (n=487). Concentrations of hydrogen in exhaled air and blood glucose were measured. Afterwards, subjects were asked to provide oral mucosa samples for genotyping and answer a questionnaire (participation rate 56%, n=274).

RESULTS: Mean hydrogen levels in exhaled air at 120min were significantly higher in the CT genotype than in the TT genotype. There was no significant difference in blood glucose levels between the two groups. Reported symptoms, with the possible exception of abdominal pain, were equally prevalent in both groups.

CONCLUSIONS: Subjects with the CT and TT genotypes, hitherto classified as lactase-persistent, differ in their physiological response to lactose intake, indicating differences in phenotype which could have clinical significance.

KEYWORDS: 13910 C/T allele; Abdominal pain; Hydrogen breath test; Oral lactose load test

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