TECHNOLOGY

Novel Genetic Mutations Associated with Lactose Persistence in African and Middle Eastern Populations.

OVERVIEW

In humans, lactose intolerance is caused by the inability to metabolize lactose, the main carbohydrate present in milk. In adults the ability to digest lactose declines rapidly after weaning because of decreasing levels of the enzyme lactase phlorozin hydrolase (LPH). LPH is predominantly expressed in the small intestine, where it hydrolyzes lactose into glucose and galactose, sugars that are easily absorbed into the bloodstream. Lactose intolerance is the most common genetic disorder affecting more than 50% of adult population worldwide. Its prevalence varies from as high as 98% in Southeast Asians to 5% in North Europeans. The symptoms associated with lactose intolerance are relatively mild and are treated by consuming a lactose-free diet. Nevertheless dietary restrictions may lead to reduction of calcium intake which may have potential clinical consequences including reduction of bone mineral mass, reduction of bone density and eventually osteoporosis and fractures in pre-menstrual women and the elderly.

Researchers at the University of Maryland have identified three novel mutations that are associated with lactase persistence (the ability to digest milk in adults), and non-persistence (lactose intolerance) in East Africans, North Africans, and some Middle Eastern populations. The researchers have demonstrated that these mutations play a role in regulating expression of the lactase phlorozin hydrolase (LPH). The three mutations could potentially be used as part of a genetic test to diagnose lactase persistence/non-persistence in individuals from African and Middle Eastern descent.

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Additional Information

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PATENT STATUS

Patent(s) pending

LICENSE STATUS

Contact OTC for licensing information

CATEGORIES

- Diagnostics

EXTERNAL RESOURCES

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