

Lactose Intolerance

Cecilia Humphrey

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Dr. Emma Emanuel

Lactose Intolerance, also known as lactose malabsorption, is a condition in which the small intestines cannot digest the lactose, or, milk sugar, in dairy products (Holmes, S. (2006). In order for lactose to be digested in the gastrointestinal tract, the enzyme lactase needs to be present. The enzyme Lactase breaks down the lactose into galactose and glucose units so that they can be absorbed into the bloodstream (Ophardt, C., & Emeritus, P. 2019, June 5). For people who are lactose intolerance, there is either a lack of abundance of lactase enzymes, or in extreme cases, none at all (Holmes, S. (2006). The lactase deficiency is called hypolactasia, while the complete absence of lactase is called alactasia (Holmes, S. (2006). Chemically, lactose is made from galactose and glucose units bonded by an acetal oxygen bridge (Ophardt, C., & Emeritus, P. 2019, June 5). There is a discrepancy in when the intolerance develops- either in childhood or later in adulthood. Within these developmental timelines is a differentiated cultural impact of lactose intolerance. Europeans are the least likely to have it, while Asians, African Americans, Africans and Caribbeans are the most likely (Definition & Facts for Lactose Intolerance. 2018). There are several theories to explore for why this cultural discrepancy occurs and why there are even variations within the same culture.

As a general overview, lactose intolerance occurs genetically in two different ways: being born with it, or development in adulthood (Definition & Facts for Lactose Intolerance 2018). The most common type of lactose intolerance is lactase non-persistence where lactase production lessens after infancy while symptoms develop in late childhood, teen years or adulthood (Definition & Facts for Lactose Intolerance 2018). This type of intolerance is dependent on the variation of the *MCM6* gene that gets inherited from parents (Lactose intolerance 2020). The *MCM6* gene is located on the arm of the 2nd chromosome at the Cytogenetic Location: 2q21.3

(MCM6 gene, 2020). The *MCM6* gene contains a specific DNA sequence for a regulatory element that controls another gene called *LCT* which holds the instructions for making the enzyme lactase (Lactose intolerance 2020). If a person, by adulthood, has not become lactose intolerant, it is because they have inherited one of the four variations of the regulatory element that affects *LCT*'s gene expression (MCM6 gene, 2020). These gene variations are autosomal dominant which means only one working copy is necessary to promote lactase production. Those without these mutations from one parent will be lactose intolerant to some degree (MCM6 gene, 2020), in fact, approximately 75% of the world's population develops lactose intolerance at some point (Mattar, R., Campos Mazo, D. F. D., & Carrilho, F. J., 2012).

The second type of lactose intolerance is less common and is called congenital lactase deficiency (congenital alactasia) which is when babies are unable to break down the lactose in breast milk and formula (Lactose intolerance 2020). CLD is believed to be an autosomal recessive trait, however there is not much known about its molecular basis (Mađry, E., Fidler, E., & Walkowiak, J. 2010). The place where cases of CLD are most commonly found is Finland where approximately 1 in every 60,000 newborn has this disease (Lactose intolerance 2020). While this sect of lactose intolerance is not as far reaching as lactase non-persistence, data suggests that the hereditary nature of the intolerance stems from an ethnicities or cultures relationship and reliance on milk and/or milk products (Mađry, E., Fidler, E., & Walkowiak, J. 2010). In Europe, lactose intolerance increases South and Eastward, reaching 70% in southern Italy and Turkey (Mađry, E., Fidler, E., & Walkowiak, J. 2010). In the caucasian population it is rare to see intolerance in children younger than five, whereas people of color as a general group have symptoms shown as early as two years old.

## References

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Disaccharides/Lactose