

Genes-4U

Lactose Intolerance : Mutation T-13910C of the Lactase gene (LCT T-13910C)

About 15 – 25 % of Caucasians and an even higher percentage of individuals of African or Far Eastern descent are affected by Lactose Intolerance (Adult-type Lactase-Non-Persistence).

In these individuals the capacity to metabolise Lactose decreases steadily from the second year after birth onwards. The failure to metabolise Lactose, the common sugar in milk, is associated with intolerance of milk and certain milk products, gastrointestinal symptoms due to enteric bacterial overgrowth, irritable bowel syndrome and diarrhea, failure to thrive and malnourishment if not diagnosed. Often Lactose intolerance is also associated with failure to absorb calcium from milk and milk products and therefore may interfere with bone development at early age or contribute to osteoporosis later in life.

The genetic basis for this common metabolic abnormality was first described in 2002 as a mutation in a regulatory region of the Lactase gene several kilobases upstream of the Lactase gene. **Further studies have substantiated the notion that the mutation at T-13910C 5' before the Lactase gene (LCT T-13910C) is practically diagnostic for this most common cause of Lactose Intolerance.**

The rare connatal form of Lactose Intolerance is not caused by this regulatory mutation but by disabling mutations in the Lactase gene itself or by its complete deletion. Also, secondary forms of Lactose Intolerance that accompany gastrointestinal disorders such as Crohn's disease and celiac disease are not caused by this mutation. However, due to its high frequency, the LCT T-13910C mutation may co-exist with such conditions and aggravate symptoms.

Classic diagnostic procedures for Lactose Intolerance include Lactose provocation tests with or without measurement of exhaled hydrogen ("Lactose Breath test"), or biochemical analyses of Lactose activity in biopsy samples obtained by gastro-duodenoscopy. In contrast, the molecular analysis of the LCT T-13910C mutation is performed on DNA from either blood or buccal samples obtained by non-invasive procedures and without any risk of provoking symptoms. Furthermore it can be performed at a point in time before Lactase activity declines and therefore in a pre-symptomatic time period. If the proband shows the mutation, either Lactose-free diet or substitution of Lactase can be instituted before any symptoms of Intolerance become evident.

References

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