

# PRIMARY HYPOLACTASIA - LITERATURE REVIEW

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## ABSTRACT

Lactose is a disaccharide present in mammalian milk and its digestion is carried out by enzyme lactase-phlorizin hydrolase (lactase). This enzyme is present in the villi of the small intestine and has the function of hydrolyzing the lactose molecule into two monosaccharides, glucose and galactose. Lactose intolerance is due to a dysfunction which occurs at level of the small intestine caused by the absence of lactase. It is defined as a clinical syndrome characterized by the presence of one or more symptoms such as abdominal pain, nausea, flatulence, abdominal distension and diarrhea that occurs after ingestion of lactose or lactose-containing products.

**KEYWORDS:** Lactose Intolerance, primary hypolactasia, malabsorption.

## 1. INTRODUÇÃO

Lactose is a disaccharide present in milk of mammals and humans in its digestion is carried by lactase-phlorizin hydrolase, more commonly known as lactase. Lactase is present in the villi of the small intestine and has the function of hydrolyzing the lactose molecule into two monosaccharides, glucose and galactose so that these can be absorbed by intestinal cells<sup>1,2,3,4</sup>.

The concentration of lactase enzyme distributed throughout the intestine varies, being a 40% lower activity in the duodenum as compared to the jejunum. The lactose malabsorption does not necessarily lead to intolerance symptoms of lactose and only with the onset of abdominal symptoms of lactose malabsorption that can characterize an individual as having lactose intolerance<sup>5</sup>.

Lactose intolerance is due to a dysfunction which occurs at the level of the small intestine caused by the absence of lactase. From the etiology of enzyme defect, lactase deficiency may be classified as a congenital, primary or secondary disorder<sup>4,6</sup>.

The congenital origin of intolerance is a rare disease

that occurs due to deficiency of the lactase jejunal babies, since lactose intolerance secondary source can be triggered by an intestinal injury, such as celiac disease, Crohn's disease and malnutrition<sup>2,3,6</sup>.

The primary source of intolerance is characterized by being permanent, and can cause premature in a congenital deficiency of lactase, while in adults can cause a deficiency of lactase<sup>6,7</sup>. The main difference between the congenital lactose intolerance and the primary adult hypolactasia is the molecular level, which, in the first there is an absence of lactase enzyme is or is truncated, whereas in the second, the enzyme lactase is normal, however there a reduction in their expression throughout life<sup>5</sup>.

The primary hypolactasia is the most common type of lactose intolerance, in which there is a reduction, genetically programmed, the concentration of lactase from two to three years of age, and that intolerance symptoms resulting from this become more evident teeth in adolescence or adulthood<sup>3,8,9</sup>.

## 2. MATERIAL AND METHODS

It is an article of literature review. Journals were selected in the main databases, as Scielo, PubMed and Lilacs. The keywords used for research include "Hi-primary polactasia", "lactose intolerance" and "primary hypolactasia Diagnostics". Then the most relevant articles were used as a basis for writing this article.

## 3. LITERATURE REVIEW

The undigested lactose when it becomes a source of substrate for the microorganisms of the colon, being fermented and converted to lactic acid, methane and hydrogen gas. The gas produced generates abdominal bloating and flatulence. Lactic acid, in turn, is an osmotically active compound such that draws water into the

intestinal lumen, resulting in diarrhea<sup>3,7</sup>.

Still occurs the production of volatile fatty acids which lead to acidification of stool, producing a pH below 5.5<sup>2</sup>. The gases are still suffering intestinal absorption and are subsequently eliminated by breathing, promoting an important diagnostic source<sup>5</sup>.

Thus, lactose intolerance is defined as a clinical syndrome characterized by the presence of one or more symptoms such as abdominal pain, nausea, flatulence, abdominal distension and diarrhea that occurs after ingestion of lactose or lactose containing products<sup>3,4,9</sup>.

Abdominal pain is usually located in periumbilical region or lower quadrant. The stools are typically bulky, frothy and watery. Importantly, that even patients with chronic diarrhea, weight loss is not often this finding<sup>5</sup>.

It is believed that lactose intolerance is also associated with systemic symptoms such as headache and dizziness, muscle and joint aches, fatigue, mouth ulcers, cardiac arrhythmias, pollakiuria, and others<sup>5,10</sup>.

Symptoms resulting from the ingestion of lactose vary with the individual, and depend on the amount of ingested lactose, the degree of deficiency of the lactase enzyme and the type of food with which the lactose has been consumed<sup>3,4</sup>.

For the diagnostic evaluation, we can cite methods that assess the absorption of lactose, as the intolerance test lactose, hydrogen breath test and exhaled carbon dioxide and genetic testing<sup>4</sup>.

Lactose intolerance constitutes intake test 25 to 50 grams lactose individual with the subsequent evaluation of their glycemic index. Fasting glucose is compared with the blood glucose curve after ingesting lactose, so that patients who absorb lactose obtains an increase of 1.4 mmol / L (25 mg / dL) or higher in glucose<sup>5,7,11</sup>.

The hydrogen breath test measures the expired concentration of hydrogen in the breath sample after ingestion of lactose. It is based on the idea of producing hydrogen from the fermentation in the colon of carbohydrates that are not absorbed. The test is considered positive when there is an increase of 20 ppm (parts per million) compared to baseline<sup>5,11</sup>. This is considered the gold standard for the diagnosis of lactose intolerance<sup>5</sup>.

Genetic testing is performed by collecting the patient's blood sample, with subsequent analysis of their DNA for the presence of polymorphisms in the LCT gene (gene of human lactase)<sup>1</sup>.

Special care must be taken with the treatment of patients lactose intolerant, because the total and definitive exclusion power lactase can cause injury to the individual's nutrition with respect to calcium, phosphorus, and vitamins, may trigger a reduction in density bone mineral and consequently lead to fractures<sup>5</sup>.

## 4. CONCLUSION

Early diagnosis and proper management of patients with lactose intolerance can lead to improvements in their quality of life.

There are several diagnostic options present in our midst and that used reliably translate into excellent tools to reach a definitive diagnosis.

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