

Characterization of Patients with Lactose Intolerance at a Gastroenterology Diagnostic Center

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Abstract

Introduction: The estimated global prevalence of lactose intolerance is 68%, while in Colombia, it reaches up to 59%. Primary lactase deficiency is the most common cause of lactose intolerance and malabsorption in adults. The lactose hydrogen breath test (HBT) is a routinely used diagnostic tool for this condition. **Methodology:** All lactose hydrogen breath tests (HBTs) conducted between August 2020 and September 2023 at the Gastropack medical center in Cartagena were reviewed. These tests were performed on patients presenting with symptoms such as abdominal pain, bloating, and diarrhea during gastroenterology consultations. Medical records were analyzed, and all clinical and sociodemographic variables were documented to characterize the population with lactose intolerance. **Results:** Out of 397 hydrogen breath tests performed between August 2020 and September 2023, 62 were identified as lactose HBTs. Of these, 24.4% were female patients, with an average age of 47 years. The most common symptoms included abdominal pain (71%), bloating (66.1%), diarrhea (30%), and flatulence (29%). Additionally, 38.7% of patients had a prior diagnosis of irritable bowel syndrome (IBS). Overall, 51.6% of the tests were positive for lactose intolerance. Among IBS patients, 44% tested positive for lactose intolerance. No statistically significant differences were found between patients with and without lactose intolerance. **Conclusion:** This study did not identify specific factors associated with lactose intolerance. Although the sample size was relatively small, performing lactose hydrogen breath tests is recommended in patients with suggestive symptoms and in those with conditions that share similar symptoms, such as IBS. This approach can help prevent unnecessary dietary restrictions that may negatively impact patients' quality of life and nutritional status.

Keywords

Lactose intolerance, abdominal pain, diarrhea, irritable bowel syndrome.

INTRODUCTION

Lactose is a disaccharide composed of D-galactose and D-glucose. It is the primary source of sugar in milk and the main carbohydrate in human milk, necessary for the proper growth of newborns⁽¹⁾. The enzyme lactase, which is required for the adequate absorption of lactose from breast milk, is present from the 8th week of gestation, with a maximum peak expression at birth⁽²⁾.

Primary lactase deficiency is the most common cause of malabsorption and lactose intolerance (LI) in adults. It is secondary to two single nucleotide polymorphisms in the lactase gene on chromosome 2q21, which leads to a genetically programmed progressive decrease in lactase synthesis and impaired lactose absorption at the enterocyte level. This creates an osmotic load in the intestinal lumen, which in turn causes osmotic diarrhea. When fermented by the gut microbiota, it leads to the formation of short-chain fatty acids,

hydrogen, carbon dioxide, and methane, resulting in symptoms such as abdominal pain, bloating, and flatulence⁽³⁾.

The terms “lactose malabsorption” and “lactose intolerance” should be differentiated. Lactose malabsorption refers to any cause of an inability to absorb lactose in the small intestine due to low activity or absence of the lactase enzyme. In contrast, lactose intolerance (LI) refers to the point at which the lactose load exceeds the residual activity of the lactase enzyme, leading to the onset of symptoms⁽¹⁾.

LI is a very common condition worldwide, with an estimated prevalence of approximately 68%⁽⁴⁾. Studies conducted in Colombia show an approximate prevalence of 59% using hydrogen breath tests following lactose loading, and up to 80% based on genotypic studies^(5,6). The prevalence of LI varies greatly depending on geographic location, ranging from 20% in Europe to as high as 100% in South Korea⁽⁴⁾. The actual prevalence of LI is difficult to estimate because significant differences exist between ethnic groups, and the variability of diagnostic methods complicates the interpretation of results.

The nonspecific nature of its symptoms and the overlap with other conditions, such as irritable bowel syndrome (IBS), can lead to underdiagnosis and prevent some patients from receiving the appropriate treatment and recommendations for managing their symptoms, which ultimately results in a decreased quality of life. Identifying risk factors can help make an early diagnosis, which can improve symptoms and enhance the quality of life for patients.

OBJECTIVE

The objective of this study was to identify the factors associated with lactose intolerance in a group of patients who attended a gastroenterology diagnostic center for symptoms of bloating, abdominal pain, and diarrhea, with or without a previous diagnosis of IBS or dyspepsia.

METHODOLOGY

A retrospective cross-sectional study was conducted, including patients over 18 years of age who visited the gastroenterology clinic for symptoms of bloating, abdominal pain, and diarrhea, with or without a prior diagnosis of IBS or dyspepsia. These patients underwent a lactulose hydrogen breath test (LHBT) between August 2020 and September 2023. All medical records were reviewed, and clinical variables such as abdominal pain, bloating, constipation, diarrhea, nausea, and flatulence were recorded, as well as sociodemographic variables, including a history of hypertension, diabetes mellitus, dyslipidemia, and smoking.

The study protocol was carried out in accordance with the principles of the Declaration of Helsinki for medical

research and Resolution 8530, which regulates clinical research in Colombia. The privacy of medical records was respected at all times. The institutional ethics committee approved the protocol, and it was considered a minimal-risk study for patients. Quantitative and qualitative variables were expressed as mean (SD) and percentages, respectively. Hypothesis testing was performed using a Student's t-test or Chi-square test, as appropriate. Statistical significance was set at $p \leq 0.05$.

Lactose load hydrogen breath test

Four weeks prior to the study, the use of antibiotics and any endoscopic procedures that could cause variations in the intestinal microbiota were suspended. 24 hours before the study, the consumption of dairy products, fruits, or fruit juices, as well as fiber, beans, peas, cabbage, leeks, onions, or garlic, was restricted. 12 hours before the procedure, smoking and chewing gum were prohibited.

Study procedure

After a 14-hour fast, each patient was given 25 g of lactose dissolved in 250 mL of water. Breath hydrogen samples were taken at 15, 30, 45, 60, 90, 120, 150, and 180 minutes, along with an assessment of symptoms at each sample collection point. The hydrogen breath test was considered positive if a level of >20 parts per million (ppm) was detected at 80 minutes after the lactose administration.

RESULTS

Out of a total of 397 hydrogen breath tests conducted between August 2020 and September 2023 at the Gastropack medical center, 62 LHBTs were identified. Of these, 24.4% were women, with an average age of 47 years. Additionally, 38.7% had a prior diagnosis of IBS. A total of 51.6% of the tests were positive, with a direct proportional increase to age, as the highest number of affected patients was found in those over 50 years old (40%) (**Table 1**). The most frequent symptoms were abdominal pain (71%), followed by abdominal distension (66.1%), diarrhea (30%), and flatulence (29%) (**Table 2**). Comorbidities found included hypertension (17%), dyslipidemia (9%), and hypothyroidism (5%) (**Table 3**).

In patients with lactose intolerance, the average age was 49 years, and 25% were female. A total of 44% (11/24) of patients with IBS were lactose intolerant. They had few comorbidities, such as hypertension, dyslipidemia, or hypothyroidism. The most frequent symptom was abdominal distension (24%), followed by abdominal pain (21%), diarrhea (12%), and flatulence (10%). When comparing

Table 1. General characteristics of the population

Variable	n = 62
Sex	
- Male	75.8% (47)
- Female	24.2% (15)
Age (SD)	47.7 (16.2)
Irritable bowel syndrome	38.7% (24)
Lactose intolerance	51.6% (32)
Age ranges in patients with lactose intolerance	
- Range between 20 and 30 years	6.3% (2)
- Range between 31 and 40 years	28.1% (9)
- Range between 41 and 50 years	21.9% (7)
- Over 50 years	40.6% (13)

Table created by the authors.

Table 2. Clinical manifestations of the population

Variable	n = 62
Abdominal pain	71% (44)
Abdominal distension	66.1% (41)
Constipation	19% (12)
Diarrhea	30.6% (19)
Emesis	1.6% (1)
Nausea	6.5% (4)
Flatulence	29% (18)

Table created by the authors.

Table 3. Personal history of the population

Variable	n = 62
Personal history	
- Arterial hypertension	17 % (11)
- Dyslipidemia	14.5% (9)
- Cardiovascular disease	4.8% (3)
- OSAHS	3.2% (2)
- Hypothyroidism	8.1% (5)
- Type 2 diabetes mellitus	1.6% (1)
- Smoking	4.8% (3)

OSAHS: obstructive sleep apnea/hypopnea syndrome. Table created by the authors.

the age of patients with and without lactose intolerance, no statistically significant difference was found. Although the symptoms of abdominal distension, diarrhea, and flatulence were more frequent in the lactose-intolerant group compared to patients without lactose intolerance, no statistically significant differences were observed (**Table 4**).

DISCUSSION

Of all the patients who underwent the hydrogen breath test, 38.7% had a history of IBS, and of the patients with lactose intolerance (LI), 34% had concomitant IBS. This finding supports the existing interrelation between these two conditions and also broadens the scope of potentially affected patients, considering that the global prevalence of IBS ranges from 9% to 23%⁽⁷⁾.

Previous studies have shown a clear relationship between LI and IBS. In a 2016 case-control study conducted in Germany, which evaluated the comorbidity burden reported in 6758 medical records of patients with and without LI, it was found that the presence of IBS was five times more common in patients with LI⁽⁸⁾. Some authors attribute these findings to the fact that many patients with LI are misdiagnosed as having IBS.

In a prospective study conducted in the Netherlands in 2001, 70 patients with a prior diagnosis of IBS were recruited, and LI was detected in 24.3% of them. There were no differences in symptoms between patients with a positive lactose tolerance test and those with a negative result. However, after following a lactose-free diet, gastrointestinal symptoms improved statistically significantly in these patients⁽⁹⁾.

Another study conducted in Italy, aimed primarily at identifying patients with LI among those with a prior diagnosis of IBS, enrolled 259 IBS patients and 108 control patients. Among the IBS patients, 79.9% tested positive for the hydrogen breath test, compared to 25% in the control group, with a statistically significant difference ($p < 0.001$). These findings highlight the importance of ruling out lactose intolerance in patients with a prior IBS diagnosis⁽¹⁰⁾.

In contrast, a study conducted in 2004 found no relationship between both conditions and suggested that the diagnosis of LI in patients with IBS may result in false positives, as IBS itself is associated with bacterial overgrowth, which can distort hydrogen breath test results. Furthermore, the continuous consumption of dairy products in intolerant patients may be linked to persistent irritation of the gastrointestinal tract, which could ultimately evolve into IBS^(11, 12).

Regarding symptoms, patients with LI more frequently presented with abdominal distension, diarrhea, flatulence, and nausea compared to those without LI, although these differences were not statistically significant. These findings are consistent with the fact that the symptoms of LI are sha-

Table 4. Characterization of patients with and without lactose intolerance

Variables	With lactose intolerance n = 32 (%)	Without lactose intolerance n = 30 (%)	p-Value
Female	25 (8)	23.3 (7)	0.87
IBS	34.4 (11)	43.3 (13)	0.46
Age (SD)	49.2 (15.8)	44.9 (16.5)	0.3
Arterial hypertension	9.4 (3)	26.7% (8)	0.07
Dyslipidemia	15.6 (5)	13.3 (4)	0.79
Hypothyroidism	12.5 (4)	3.3 (1)	0.18
Smoking	9.4 (3)	0 (0)	0.08
Abdominal pain	65.5 (21)	76.7 (23)	0.33
Abdominal distension	75 (24)	56.7 (17)	0.12
Constipation	12.5 (4)	26.7 (8)	0.15
Diarrhea	37.5 (12)	23.3 (7)	0.22
Nausea	9.4 (3)	3.4 (1)	0.35
Flatulence	31.3 (10)	26.7 (8)	0.69

SD: standard deviation. Table created by the authors.

red by many other conditions, including IBS, inflammatory bowel disease, celiac disease, and even malignant gastrointestinal conditions⁽¹³⁾.

In 2010, a systematic review was conducted to evaluate the diagnostic value of the most common gastrointestinal symptoms of LI (diarrhea, abdominal pain, abdominal distension, flatulence, and constipation), and the results were highly heterogeneous. For example, diarrhea had a sensitivity ranging from 30% to 80%, and specificity ranging from 32% to 84%. Similar trends were observed with the other symptoms, which supports the assertion that symptoms alone are insufficient for diagnosis⁽¹⁴⁾.

The diagnostic value of self-reported lactose intolerance remains a topic of debate. On one hand, many more individuals seem to attribute their symptoms to lactose intake than what objective tests are able to confirm. On the other hand, some patients have a positive hydrogen breath test for lactose intolerance, but they have not recognized the association between dairy consumption and the presence of symptoms^(15,16).

There are different tests to evaluate lactose intolerance. In our study, we used the hydrogen breath test, which has the advantage of being widely available and having an adequate diagnostic performance, with a sensitivity of 78% and specificity of 98%⁽¹⁷⁾.

The importance of an accurate diagnosis lies in avoiding unnecessary restrictions on dairy foods in the diet, as this could be associated with demineralization and subsequent reduction in bone density⁽¹⁸⁾. These phenomena have previously been demonstrated in children aged 3 to 10 years⁽¹⁹⁾. In adult patients, a prospective study in postmenopausal Korean women evaluated the consumption of dairy products, including milk and yogurt, and the incidence of osteoporosis. The study found that women with a high intake of these dairy products had a 48% lower risk of osteoporosis in the radial bone compared to those who did not consume them⁽²⁰⁾.

The impact of dairy consumption on cardiovascular risk has also been evaluated. A meta-analysis of 22 studies found a 12% reduction in the total risk of cardiovascular disease and a 13% reduction in the risk of presenting acute coronary syndrome with the consumption of dairy products⁽²¹⁾.

No factors associated with LI were identified, so it is recommended to rule it out in patients with compatible symptoms, even if these symptoms are similar to those found in other pathologies. This helps avoid unnecessary dietary restrictions that could affect patients' quality of life and nutritional status.

This study has limitations, including its retrospective, single-center design, limited to a sample of patients from the gastroenterology clinic. Additionally, the symptoms taken

from the medical records may have a bias of underreporting of clinical manifestations in some of the cases studied.

CONCLUSION

In the present study, no factors associated with lactose intolerance were identified. Although the number of

patients is not very large, it is recommended to perform the lactose overload hydrogen breath test in patients with symptoms suggestive of this condition, as well as in diseases with similar symptoms, such as IBS. This will help avoid unnecessary dietary restrictions that could affect the quality of life and nutritional status of the affected patients.

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