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MODERN METHODS FOR DIAGNOSING LACTASE DEFICIENCY

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Abstract. Lactase deficiency (LD) refers to a disruption of lactase enzyme activity or its complete absence, which in turn reduces the body's ability to digest lactose. This thesis analyzes modern diagnostic methods for identifying lactase deficiency, including genetic tests, breath tests, and various laboratory analyses. The use of these methods provides opportunities for effective treatment and dietary planning for patients.

Keywords: Lactase Deficiency Diagnosis, Lactose Tolerance Testing, Biomarkers of Lactase Deficiency, Symptom-Based Diagnosis, Food Challenge Method

Introduction. Lactase is the only enzyme that breaks down lactose, a sugar found in milk. Lactase deficiency, including lactose intolerance or malabsorption, is a widespread metabolic disorder affecting individuals globally. It leads to an inability or reduced ability to digest milk, causing symptoms such as pain, bloating, diarrhea, discomfort, and deficiencies in vitamins and minerals. Several diagnostic methods exist for detecting lactase deficiency, each with its advantages and limitations [1,2].

Objective. The aim of this thesis is to analyze the modern diagnostic methods used for diagnosing lactase deficiency. The focus is on the accuracy, safety, and selection of the most effective and convenient options for patients [2].

Results. Among the modern diagnostic methods, the most commonly used are:

- 1. **Breath Test (H₂ breath tests):** In this test, the level of hydrogen in the breath is measured after the consumption of dairy products. An increase in hydrogen levels is indicative of lactase deficiency [1,2].
- 2. **Genetic Tests:** In this method, the patient's DNA is analyzed to identify genetic mutations related to lactase enzyme production. The advantage of this test is its precision and reliability [2,3].
- 3. **Biochemical Tests:** Biological markers in milk or urine are analyzed to assess lactase activity [1].
- 4. **Biopsy and Histological Analysis:** In some cases, a biopsy of enterocytes from the intestine may be analyzed, and microscopic examinations are conducted. This method provides accurate and detailed results [1,2,3].

Conclusion. Modern diagnostic methods for detecting lactase deficiency complement each other, with each offering unique advantages. Breath tests are the most commonly used

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and simple method, while genetic tests provide more precise and comprehensive results. Effective diagnosis of lactase deficiency-related issues improves patients' daily lives and enables appropriate treatment to be provided.

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