2. BIOCHEMICAL ASPECTS OF MALABSORPTION SYNDROMES

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Introduction. Malabsorption affects millions of people worldwide. Prevalence of coeliac disease in the general population ranges from 0,5% to 2%, with an average of about 1%. At present over 100,000 people suffer from cystic fibrosis worldwide. Malabsorption can be caused by many different diseases and may lead to a wide spectrum of clinical signs, symptoms and biochemical findings including vitamin, mineral and macronutrient deficiency syndromes.

Aim of study. Malabsorption is a clinical term that refers to the impaired absorption of nutrients. It encompasses defects that occur during the digestion and absorption of food nutrients by, and infections of, the gastrointestinal tract.

Methods and materials. In the Pubmed and Google Scholar databases, we selected and analyzed 30 articles with embedded keywords: “Malabsorption syndromes”, “Coeliac Disease”, “Cystic Fibrosis”, etc.

Results. The absorption of nutrients occurs in three distinct stages: luminal, mucosal, and postabsorptive. Malabsorption syndromes can be classified depending on which of these three stages is affected. There are numerous causes of malabsorption, including lactose intolerance, inflammatory bowel diseases, cystic fibrosis, short bowel syndrome, and others. Therefore, it is important to thoroughly examine the family history. From a biochemical perspective, malabsorption syndromes can be classified based on which nutrients are not assimilated: proteins, fats, carbohydrates, or micronutrients. Malabsorption syndromes are manifested by both biochemical abnormalities (anemia, hypoalbuminemia, dyslipidemia, vitamin deficiencies, etc.) and a clinical triad, including chronic diarrhea, abdominal distension, and developmental and/or growth delay. Celiac disease should be considered a primary food-related condition with lifelong consequences for affected individuals. The specific role of HLA-DQA1 and HLA-DQB1 genes in presenting gluten peptides as antigens makes the MHC-HLA locus a crucial genetic factor in the development of celiac disease. Nutrition is closely linked to the disease outcome, and malnutrition in cystic fibrosis typically results in a chronic negative energy balance, leading to malabsorption due to pancreatic enzyme insufficiency.

Conclusion. Nutrient absorption can be disrupted by numerous pathophysiological conditions, including genetic disorders, such as those seen in celiac disease. Understanding the biochemical mechanisms of malabsorption syndromes is crucial for the diagnosis and management of these pathologies.