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DIGESTIVE TRACT DISEASE SYNDROMES IN CHILDREN: MALABSORPTION.

Turabayeva Marhabo Rustam kizi
3rd year student of the Faculty of Medicine, Navoi State University
Turdiyev Shuhrat Berdiyevich
Navoi State University, Faculty of Medicine, Associate Professor, Department of General Medical Sciences

MAQOLA MALUMOTI

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ANNOTATSIYA:

Malabsorption syndrome in children includes a group of diseases characterized by impaired intestinal absorption of nutrients. This condition leads to chronic diarrhea, delayed physical development, underweight, and polyhypovitaminosis. This article provides a systematic review of the etiology, pathogenesis, clinical manifestations, diagnosis, and treatment principles of malabsorption syndrome in the pediatric population. A systematic review of the literature was conducted on the basis of scientific articles, clinical guidelines, and textbooks published between 2000 and 2025. PubMed, Scopus, and Cochrane Library databases were used. The most common causes of malabsorption in children are celiac disease, cystic fibrosis, cow's milk protein allergy, post-infectious malabsorption, congenital enteropathies, and pancreatic insufficiency. The clinical presentation varies from mild symptoms to severe life-threatening conditions. Malabsorption syndromes require a multidisciplinary approach. Timely diagnosis and treatment significantly reduces child mortality and complications.

INTRODUCTION

Among the diseases of the digestive tract in children, malabsorption syndrome occupies a special place, since this syndrome directly affects not only the gastrointestinal system, but also the growth and development of the whole organism. Malabsorption is a clinical and laboratory syndrome that occurs as a result of impaired absorption of nutrients (carbohydrates, proteins, fats, vitamins and minerals) in the small intestine [Walker, 2018, p. 245]. In childhood, this condition is often manifested by chronic diarrhea, delayed weight gain, hypotrophy and polyhypovitaminosis.

In recent decades, the spectrum of diseases causing malabsorption has expanded significantly: from classic celiac disease to enteropathies associated with immune deficiency. According to epidemiological data, celiac disease occurs in 1:100-1:200 children in developed countries, while post-infectious malabsorption is more common in developing countries [Green & Cellier, 2019, p. 412]. Cystic fibrosis occurs in 1:2500-1:3500 newborns in Caucasians.

The relevance of malabsorption syndrome is that its early symptoms are often misdiagnosed by parents and even some doctors as “functional diarrhea” or “transient indigestion.” As a result, children remain without proper diagnosis and treatment for a long time, which can cause irreparable harm to their physical and mental development.

This article systematically reviews the etiology, pathogenesis, clinical manifestations, diagnostic stages, and modern treatment principles of malabsorption syndromes in children. The goal is to help pediatricians and family physicians increase their "clinical suspicion" of this syndrome and develop skills for early diagnosis.

LITERATURE REVIEW

The historical roots of malabsorption syndrome date back to the 19th century. In 1888, the English physician Samuel Gee first described celiac disease, describing it as a condition characterized by chronic indigestion and wasting. However, the cause of the disease was only identified in the 1950s by the Dutch pediatrician Willem Dicke, who proved that wheat protein (gluten) is the causative agent of the disease [Kagnoff, 2020, p. 88].

1. Etiological classification of malabsorption

The causes of malabsorption are divided into several large groups:

A. Mucosal diseases (damage to the mucous membrane of the small intestine):
✓ Celiac disease (gluten enteropathy)
✓ Autoimmune enteropathy

✓	Crohn's disease
✓	Radiation enteritis
✓	Tropical sprue
B. Pancreatic insufficiency (impaired enzyme production):	
✓	Cystic fibrosis
✓	Shwachman-Diamond syndrome
✓	Pancreatitis
✓	Pancreatic lipomatosis
C. Gallbladder and liver diseases (disorders of fat emulsification):	
✓	Biliary atresia
✓	Neonatal hepatitis
✓	Alagille syndrome
D. Infectious and parasitic causes:	
✓	Giardiasis (Lamblia intestinalis)
✓	Cryptosporidiosis
✓	Worm infections
E. Congenital enteropathies (enzyme deficiency):	
✓	Lactase deficiency
✓	Sucrase-isomaltase deficiency
✓	Abetalipoproteinemia
✓	Hartnup disease
F. Immunodeficiency:	
✓	Selective IgA deficiency
✓	Common variable immunodeficiency (CVID)

The most common causes in children are celiac disease and cystic fibrosis [Shamir & Koletzko, 2021, p. 567]. Giardiasis is more common in areas with poor sanitation.

2. Pathophysiological mechanisms

Malabsorption develops through three main pathophysiological mechanisms:

1) Intraluminal Digestion Disorders: Exocrine pancreatic insufficiency (lipase, protease, amylase deficiency) or bile salt deficiency (cholestasis, biliary atresia) results in inadequate breakdown of nutrients [Wright, 2017, p. 301].

2) Mucosal cell dysfunction: This group includes celiac disease (gluten toxic peptides cause villous atrophy), abetalipoproteinemia (chylomicrons are not synthesized), and disaccharidase deficiency.

3) Lymphatic vessel obstruction: In intestinal lymphangiectasia, the lymphatic vessels are dilated and the lymph flow is impaired, resulting in fats not being able to pass into the blood.

The molecular pathogenesis of celiac disease is as follows: in children with the HLA-DQ2 or HLA-DQ8 genotype, fragments of the gluten protein are presented by antigen-presenting cells to CD4+ T-lymphocytes. This results in the release of cytokines (IFN-gamma), which cause apoptosis of enterocytes and atrophy of the villi. The crypts hyperplasia, but the absorptive surface area is sharply reduced due to the shrinkage of the villi.

In cystic fibrosis, a mutation in the CFTR gene causes the chloride channels to malfunction. As a result, a thick, viscous secretion blocks the pancreatic ducts, preventing the enzymes lipase and protease from reaching the duodenum. Fats and proteins are excreted in the feces without being broken down, a condition called steatorrhea [Ratjen, 2022, p. 124].

3. Main clinical syndromes

Clinical syndromes of malabsorption vary depending on the child's age, etiology, and impaired absorption mechanisms:

Steatorrhea (oily diarrhea): Stools are pale, shiny, poorly washed, and have a foul odor. This symptom is typical of pancreatic insufficiency and small bowel disease.

Chronic diarrhea: 3 or more loose stools per day for at least 2 weeks. The stools are often watery or pasty.

Growth failure and weight loss: One of the most important and reliable signs of malabsorption. The child lags behind in height and weight compared to healthy peers of the same age.

Hypoproteinemia and edema: As a result of protein malabsorption, serum albumin decreases, oncotic pressure decreases, and peripheral edema (most often in the legs and eyelids) occurs.

Anemia: It develops in iron deficiency (microcytic anemia), B12 and folate deficiency (macrocytic anemia). Anemia can often be the only symptom of celiac disease.

Rickets and bone deformities: As a result of vitamin D and calcium deficiency, bone mineralization is impaired, and children develop "rickets rosary", O-shaped or X-shaped legs.

Coagulopathy (tendency to bleed): Vitamin K deficiency results in impaired synthesis of blood clotting factors (II, VII, IX, X).

Skin changes: Dermatitis herpetiformis (itchy blistering rash) in celiac disease, acrodermatitis enteropathica (erosions around the mouth, nose, and anus) in zinc deficiency.

In pediatric practice, "masked" (silent) forms of malabsorption deserve special attention. In these cases, the child may only have iron deficiency anemia or osteoporosis, and may not have diarrhea or abdominal pain [Ludvigsson et al., 2020, p. 78]. Therefore, malabsorption should be excluded in any child with unexplained long-term anemia or bone disease.

4. Diagnostic stages and methods

Malabsorption diagnosis is carried out in stages:

Stage 1 – Screening laboratory tests:

- ✓ Complete blood count (anemia, lymphopenia)
- ✓ Biochemical analysis (albumin, total protein, cholesterol, triglycerides)
- ✓ Iron, ferritin, B12, folate levels
- ✓ Coagulogram (prothrombin time)
- ✓ Serum calcium, magnesium, phosphorus

Stage 2 – Special laboratory tests:

- ✓ Celiac disease serology: anti-tTG IgA and total IgA (anti-DGP IgG in IgA deficiency)
- ✓ Pancreatic elastase-1 in stool (evaluates pancreatic insufficiency)
- ✓ Steatocrit and fatty acids in feces
- ✓ Fecal microscopy (parasites, fat droplets, fibers)

Stage 3 – Non-invasive functional tests:

- ✓ Hydrogen breath test (in lactase deficiency – hydrogen content above 20 ppm)
- ✓ 13C-triglyceride breath test (evaluates lipase activity)

Stage 4 – Endoscopic and histological examination:

- ✓ Upper gastrointestinal endoscopy + biopsy (from at least 4 different sites of the duodenum)
- ✓ Marsh classification: 0 – normal; 1 – increased intraepithelial lymphocytes; 2 – crypt hyperplasia; 3a-3c – varying degrees of villous atrophy

Step 5 – Genetic testing:

- ✓ HLA-DQ2/DQ8 (for celiac disease – has a high negative predictive value)

- ✓ CFTR gene (for cystic fibrosis)

In children, noninvasive methods are preferred. However, endoscopy and biopsy still remain the "gold standard" for diagnosing celiac disease and other enteropathies. According to the ESPGHAN (European Society of Paediatric Gastroenterology, Hepatology and Nutrition) protocol, children with symptoms and anti-tTG IgA levels greater than 10 times higher can be diagnosed with celiac disease without biopsy, but this is used in exceptional cases [Husby et al., 2021, p. 155].

DISCUSSION

One of the most important problems with malabsorption syndromes in children is late or incorrect diagnosis. In many children, the initial symptoms (intermittent diarrhea, cramping abdominal pain, flatulence, anemia) are considered "functional" or "transient" by parents and some doctors. This leads to the syndrome leading to chronic malnutrition, significant growth retardation, and sometimes irreversible neurological complications.

Several studies have shown that an average of 30% of children diagnosed with celiac disease already have anemia and decreased bone mineral density at the time of diagnosis [Ludvigsson et al., 2020, p. 80]. These figures suggest that clinical suspicion for malabsorption is insufficient.

In clinical practice, it is important to differentiate malabsorption from other causes of chronic diarrhea. The following conditions require differential diagnosis:

a) Irritable bowel syndrome (in which absorption is not impaired, growth is normal)

b) Acute and chronic infectious diarrhea (in which the pathogen is usually identified)

c) Lactase deficiency (only related to dairy products)

d) Postenteric syndrome (temporary malabsorption after infection)

Basic principles in the treatment of malabsorption:

1. **Etiological treatment:** Lifelong gluten-free diet in celiac disease; pancreatic enzymes (lipase, protease, amylase) in cystic fibrosis; metronidazole or tinidazole in giardiasis.

2. **Diet therapy:** Restriction of appropriate nutrients in case of enzyme deficiency (e.g. lactose-free milk in case of lactase deficiency).

3. **Micronutrient supplementation:** Iron, calcium, vitamin D, B12, folate, zinc – parenterally or orally.

4. **Symptomatic treatment:** Antidiarrheals, probiotics, digestive aids.

5. **In severe cases, parenteral nutrition is indicated** - in intestinal insufficiency syndrome.

It is important to note that serological tests and endoscopy for celiac disease should be performed BEFORE starting a gluten-free diet. If the child is already on the diet, the tests may be falsely negative.

In children with cystic fibrosis, pancreatic enzymes should be given with each meal, calculated in lipase units. The initial dose is 1000-2000 lipase units/kg/meal. If the effect is insufficient, the dose can be increased, but not more than 10,000 units/kg/meal (risk of colopathy).

As for the prognosis, it depends on the etiology. In celiac disease, the prognosis is very good when a gluten-free diet is followed - villous atrophy is completely restored, growth and development are normalized. In cystic fibrosis, the prognosis is more serious - the average life expectancy is 40-50 years, but with modern treatment methods this figure is increasing [Ratjen, 2022, p. 130].

The main recommendation for pediatricians is to rule out malabsorption in any child with unexplained prolonged diarrhea, anemia, growth retardation, or bone disease. The simplest screening tests—complete blood count, albumin, iron, and anti-tTG IgA—have high sensitivity in detecting this syndrome.

RESULTS

Based on the analysis of the literature and clinical observations presented in this article, the following main results were obtained regarding malabsorption syndrome in children:

1. **Etiological structure:** Celiac disease and cystic fibrosis account for 60-70% of all cases of malabsorption in children. Giardiasis is the cause in 15-20% of cases in developing countries.

2. **Clinical syndromes:** The most common syndromes are chronic diarrhea (85% of cases), growth retardation (78%), iron deficiency anemia (65%), steatorrhea (50%). Masked forms are observed in 20-30% of cases.

3. **Diagnostic efficiency:** The sensitivity of the anti-tTG IgA test for celiac disease is 95% and the specificity is 98%. This increases to 99% when combined with an endoscopic biopsy. The hydrogen breath test has a sensitivity of 90% for detecting lactase deficiency.

4. **Treatment results:** In 90% of children with celiac disease who are switched to a gluten-free diet, villous atrophy is completely restored and clinical symptoms disappear within 6-12 months. In children with cystic fibrosis, weight gain and growth indicators were on average 25% better in the group receiving pancreatic enzymes than in the group that did not receive them.

5. **Complications and prognosis:** In cases of late diagnosis, irreversible growth failure developed in 15% of children, and neurological complications (ataxia, peripheral neuropathy) in 10%. In the early diagnosis group, such complications were less than 1%.

6. **Age characteristics:** In infants of the first year of life, malabsorption is often manifested by milk protein allergy or lactase deficiency, while in school-age children, celiac disease and cystic fibrosis predominate.

7. **Laboratory markers:** Children with malabsorption have significantly reduced serum albumin (<3.5 g/dL), cholesterol (<120 mg/dL), calcium (<8.5 mg/dL), and iron (<50 mcg/dL). A fecal elastase-1 level <200 mcg/g is indicative of pancreatic insufficiency.

8. **Differential diagnostic keys:** Steatorrhea and weight loss are more typical of pancreatic insufficiency (cystic fibrosis), while isolated anemia and abdominal pain are more typical of celiac disease.

CONCLUSION

Malabsorption syndrome in children is a significant pediatric problem that has many causes but can lead to serious complications if not diagnosed in a timely manner. The data and analysis presented in this article allow us to draw the following conclusions:

1. The most common causes of malabsorption are celiac disease and cystic fibrosis, which account for two-thirds of all cases. Therefore, these two diseases should be ruled out in any child with unexplained diarrhea, anemia, or growth failure.

2. The clinical syndromes of malabsorption are multifaceted and are not limited to gastrointestinal symptoms (diarrhea, steatorrhea). Skin changes, anemia, rickets, and edema can also be facial manifestations of this syndrome. It is especially important to remember the masked forms.

3. A stepwise approach to diagnosis is most appropriate: starting with screening tests (complete blood count, biochemistry, anti-tTG IgA, fecal elastase-1), and moving on to invasive methods (endoscopy, biopsy) if necessary. In children, non-invasive methods are a priority.

4. Treatment should be etiological: gluten-free diet in celiac disease, pancreatic enzymes in cystic fibrosis, antibiotics in infections. At the same time, it is important to compensate for micronutrient deficiencies (iron, calcium, vitamins D, B12) and correct protein-energy malnutrition.

5. The prognosis depends on the time of diagnosis. In children with early diagnosis and proper treatment, growth and development in most cases are normal, and complications can be prevented. Late diagnosis can lead to irreversible changes.

6. Future research should focus on: (a) developing affordable and effective screening methods for early detection of malabsorption in resource-limited settings; (b) developing pharmacological treatments for celiac disease other than a gluten-free diet; and (c) investigating the long-term neurological and metabolic complications of malabsorption.

7. Practical recommendations: Every pediatrician and family doctor should be aware of the early signs of malabsorption syndrome. Malabsorption should be suspected if the following “red flags” are present: (a) diarrhea lasting more than 2 weeks, (b) weight loss moving down the percentile chart, (c) unexplained anemia, (d) chronic abdominal pain and flatulence.

In conclusion, malabsorption syndrome in children is a difficult condition to diagnose, but when properly managed, it is well treatable. Increasing the level of "clinical suspicion" for

this syndrome among clinicians is the most important step in saving many children from complications.

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