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Early-onset inflammatory bowel disease in children: diagnostic dilemmas and management advances.

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Introduction

Inflammatory Bowel Disease (IBD), comprising Crohn's disease (CD), ulcerative colitis (UC), and IBD-unclassified (IBD-U), is increasingly recognized in the pediatric population. Early-onset IBD, particularly very early-onset IBD (VEO-IBD), diagnosed before 6 years of age, is now being identified more frequently due to better awareness, improved diagnostic tools, and evolving genetic research. Children with EO-IBD often have more extensive colonic involvement, rapid progression, and a higher likelihood of growth failure, malnutrition, and extraintestinal manifestations. This makes early diagnosis and individualized management strategies crucial to preserving quality of life and reducing long-term complications.[1].

Diagnostic dilemmas in Early-Onset Inflammatory Bowel Disease (EO-IBD) often arise due to atypical presentations, particularly in children under six years of age, where symptoms can significantly differ from the classic adult patterns. Common features include chronic diarrhea, growth retardation or failure to thrive, rectal bleeding, abdominal pain, and perianal disease (especially in Crohn's disease). However, these clinical manifestations frequently overlap with other pediatric conditions such as infections, food allergies, immunodeficiencies, or monogenic diseases, making the diagnostic process more complex and challenging [2].

Very early-onset inflammatory bowel disease (VEO-IBD) can be mistaken for several overlapping conditions, including primary immunodeficiencies such as chronic granulomatous disease (CGD) and Wiskott-Aldrich syndrome, allergic colitis, infectious colitis, and Hirschsprung-associated enterocolitis. Due to the clinical similarities among these conditions, a multidisciplinary approach is essential for accurate diagnosis and management, often requiring the collaboration of pediatric gastroenterologists, immunologists, and geneticists [3].

Recent genomic studies have identified over 50 genes associated with monogenic forms of inflammatory bowel disease (IBD), particularly affecting infants and toddlers. Mutations in genes such as IL10, FOXP3, TTC7A, and NOD2 can lead to syndromic IBD or severe enterocolitis that mimics IBD. In these cases, whole exome sequencing (WES) has emerged as a valuable diagnostic tool, especially for identifying underlying genetic causes in very young children presenting with refractory o r atypical forms of the disease [4].

Management of Early-Onset Inflammatory Bowel Disease (EO-IBD) involves a multi-faceted approach aimed at inducing and maintaining remission, supporting growth, and preventing complications. Medical therapy includes aminosalicylates (5-ASA) for mild to moderate ulcerative colitis, corticosteroids for induction only due to side effects, and immunomodulators like azathioprine and methotrexate for steroid-sparing effects. Biologic agents, particularly anti-TNF therapies such as infliximab and adalimumab, are key for moderate-to-severe cases and fistulizing Crohn's disease, with newer agents like ustekinumab (anti-IL12/23) and vedolizumab (anti-integrin) currently being explored in pediatric trials. The use of biologics earlier in the disease

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course, especially in patients with extensive disease or poor prognosis, reflects a top-down treatment strategy. Nutritional therapy, notably Exclusive Enteral Nutrition (EEN), is as effective as steroids for inducing remission in pediatric Crohn's disease and also supports growth [5].

Conclusion

Early-onset IBD in children presents diagnostic and management challenges far beyond those seen in adults. Atypical presentations, the potential for monogenic disease, and a more aggressive disease course require a comprehensive, multidisciplinary, and personalized approach. Advancements in genetics, biologics, and nutritional therapy have significantly improved disease control and outcomes. However, early detection, psychosocial support, and long-term follow-up remain pillars of effective pediatric IBD care.

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