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CHALLENGES IN THE EARLY DIAGNOSIS OF EARLY-ONSET COLORECTAL CANCER: AN INTEGRATIVE REVIEW

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ABSTRACT

Colorectal cancer has shown a progressive increase in incidence among adults under 50 years of age, characterized by aggressive biological behavior and delayed diagnosis. This study aimed to analyze the aspects related to the early diagnosis of colorectal cancer in young adults, identifying the main factors that hinder its timely detection. To this end, an integrative literature review was conducted through a systematized search in the PubMed (MEDLINE) database in May 2026, selecting 17 articles published between 2016 and 2026 in English, Portuguese, and Spanish. The results revealed that low clinical suspicion, the non-specific nature of early symptoms—frequently misattributed to benign conditions—and the historical exclusion of this age group from systematic screening programs are the primary drivers of diagnostic delay. This scenario culminates in the identification of the disease at advanced stages, requiring invasive treatments and increasing both psychosocial impact and morbidity and mortality during product life phases. In conclusion, addressing this neoplasm requires a shift from traditional age-based screening strategies toward individualized genetic and molecular risk assessments. Expanding public awareness and training primary care networks to recognize early warning signs are vital steps to improve the prognosis and quality of life for young patients.

Keywords: Colorectal neoplasms, Early diagnosis, Genetic predisposition, Mass screening, Young adult.

INTRODUCTION

The Colorectal cancer (CRC), traditionally associated with individuals over 50 years of age, has shown a progressive increase in incidence among young adults.[1] As screening is generally targeted at older age groups, younger patients tend not to undergo early diagnostic evaluation, which contributes to delayed diagnoses.[2,3]

Furthermore, the initial signs and symptoms of CRC, such as rectal bleeding, abdominal pain, changes in bowel habits, diarrhea, weight loss, and iron-deficiency anemia, are often nonspecific and may be mistaken for benign conditions, such as hemorrhoids and irritable bowel syndrome, contributing to a lack of further investigation and diagnostic delays.[1,4]

In this context, despite the presence of these warning signs, many young adults experience delays between symptom onset and diagnostic testing, influenced by patient-related factors, access to healthcare services,

and low clinical suspicion.[2,4]

Therefore, it is essential to increase clinical suspicion and consider CRC in the differential diagnosis of young adults, particularly in the presence of persistent symptoms, genetic predisposition, or associated hereditary syndromes.[4,5]

Thus, this integrative review is justified by the need to synthesize evidence on the factors that hinder the early diagnosis of CRC in young adults, contributing to greater clinical recognition.[6]

OBJECTIVES

To analyze aspects related to the early diagnosis of colorectal cancer in young adults.

METHODS

The present study is an integrative literature review with a qualitative approach. To this end, a systematic search for publications relevant to the topic was conducted, taking into account the proposed objectives.

For this review, a literature search was conducted in May 2026 using the PubMed (MEDLINE) database to obtain consistent and relevant search results, allowing for a thorough analysis and avoiding inconsistencies. The following descriptors available in the Health Sciences Descriptors (DeCS) were combined using the Boolean operators AND and OR: "Colorectal Neoplasms," "Colorectal Cancer," "Diagnosis," and "Young Adult."

The inclusion criteria comprised articles published in English, Portuguese, or Spanish, available in full text free of charge, and published within the previous ten years (2016–2026).

Incomplete studies and those not relevant to the topic were excluded, including studies focusing on other age groups, other types of cancer, or those that did not address early diagnosis.

The initial search yielded 67 publications. After applying the inclusion criteria, 39 articles remained. Following the application of the exclusion criteria, 26 articles remained. These were screened through a careful review of their titles and abstracts. After full-text assessment to determine their relevance to the topic, the final sample consisted of 17 articles, which formed the basis for the analysis and discussion presented in this study.

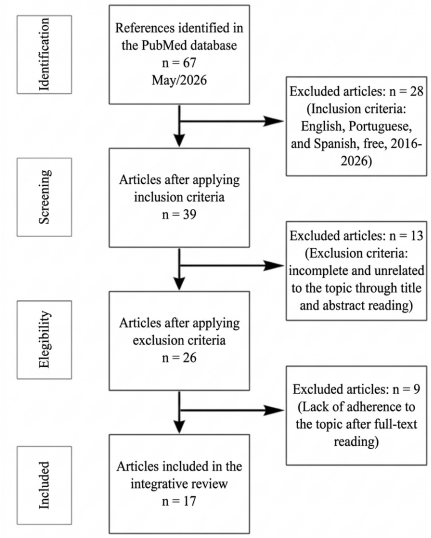


Figure 1: Methodology Flowchart.
Source: Prepared by the authors, 2026.

RESULTS AND DISCUSSION

The epidemiological landscape of colorectal cancer (CRC) has undergone a significant shift, characterized by a progressive increase in incidence among adults younger than 50 years.[7] This increase, observed globally in regions such as the Americas and East Asia,[8,9] contrasts with declining rates among older populations—who have benefited from established screening programs—suggesting that early-onset colorectal cancer (EOCRC) constitutes a distinct clinical and biological entity rather than merely an earlier manifestation of the disease typically seen in older adults.[6] Data analysis reveals that this increase is particularly pronounced in tumors located in the distal colon and rectum,[10] frequently associated with highly aggressive phenotypes, such as mucinous histology and the presence of signet-ring cells.[11]

A detailed characterization of the selected articles, including authorship, study design, and the main outcomes associated with barriers to early diagnosis, is summarized in Table 1.

Table 1: Characteristics of the Studies Included in the Integrative Review on the Challenges of Early Diagnosis of Colorectal Cancer in Young Adults

Author and year	Study design	Main findings related to diagnostic challenges
Done e Fang (2021)	Literature/Narrative Review	Global increase in early-onset colorectal cancer (EOCRC), particularly in the distal colon and rectum, associated with delayed diagnosis and gaps in screening.
Castelo et al. (2023)	Population-Based Cohort Study	Directly compares time to diagnosis between young and older adults, demonstrating significant diagnostic delays in younger patients.
Castelo et al. (2022)	Systematic Review	Synthesizes evidence on diagnostic delays, incorrect initial screening, and poorer oncological outcomes in young adults with CRC.
You et al. (2020)	Clinical/Practice Review	Addresses the prolonged interval between symptom onset and diagnosis in adolescents and young adults due to the nonspecific nature of symptoms.
Shaikh et al. (2018)	Epidemiological/Clinical Study	Demonstrates low adherence to genetic testing guidelines for dMMR in young patients, highlighting deficiencies in hereditary risk management.
Campos (2017)	Literature Review/Opinion Article	Highlights the clinical challenges of EOCRC due to biological aggressiveness (mucinous histology) and low initial clinical suspicion in young patients.

Patel e Patil (2022)	Opinion/Perspective Article	Warns of concerning trends in the increasing incidence of EOCRC and the urgent need to make strictly age-based screening criteria more flexible.
Ueno et al. (2023)	Narrative/Epidemiological Review	Discusses the epidemiological landscape in Japan, emphasizing barriers to healthcare access and the historical exclusion of young adults from screening programs.
Iberri Jaime et al. (2025)	Descriptive Cross-Sectional Study	Describes an emerging clinical profile at a referral hospital, confirming the predominance of metastatic disease at diagnosis due to delayed clinical suspicion.
Park et al. (2022)	Retrospective Study	Analyzes the clinicopathological characteristics of EOCRC, demonstrating a higher prevalence of poorly differentiated tumors and advanced-stage disease.
López-Basave et al. (2023)	Cross-Sectional/Retrospective Study	Highlights aggressive tumor characteristics and diagnoses at advanced stages (III and IV) in patients younger than 30 years.
Ansari et al. (2024)	Case Report	Illustrates the histopathological complexities and diagnostic challenges of familial adenomatous polyposis (FAP) at young ages.
Salem et al. (2021)	Retrospective Cohort Study	Reveals the critical impact of sociodemographic disparities and health insurance status on overall survival among patients with EOCRC.
Memon et al. (2019)	Case Report	Presents a case of rectal adenocarcinoma in a young patient whose initial sign (anemia) was initially underestimated or detected incidentally.
Holowatyj et al. (2019)	Clinical and Molecular Study	Associates mismatch repair deficiency (dMMR) and biological aggressiveness (signet-ring cells) with the molecular profile of younger patients.
Miller et al. (2021)	Cohort/Survivorship Study	Evaluates the long-term psychosocial impact and quality of life of young CRC survivors, both worsened by delayed diagnosis.
Chang et al. (2021)	Database-Based Epidemiological Study	Demonstrates disparities in EOCRC incidence according to race and ethnicity, highlighting structural inequalities in healthcare.

Source: Prepared by the authors, 2026.
Note: CRC = Colorectal Cancer; EOCRC = Early-Onset Colorectal Cancer; dMMR = Deficient DNA Mismatch Repair; FAP = Familial Adenomatous Polyposis.

Unlike the classical form of the disease, early-onset colorectal cancer (EOCRC) exhibits more invasive biological behavior, with a higher prevalence of poorly differentiated tumors at diagnosis.[10] The etiology of this phenomenon is recognized as multifactorial, involving a complex interaction between genetic predisposition and modifiable risk factors. Although hereditary syndromes, such as Lynch syndrome and familial adenomatous polyposis, are disproportionately relevant in cases diagnosed at very young ages,[12] most cases are sporadic and strongly influenced by substantial changes in modern lifestyle.[1] Increasing obesity rates, physical inactivity, and dietary patterns centered on ultra-processed foods and red meat have been implicated as drivers of early carcinogenesis,[8] possibly mediated by chronic inflammatory changes in the intestinal microbiota.[7]

Furthermore, the epidemiological profile is influenced by profound sociodemographic disparities that shape disease prognosis. Variables such as ethnicity and socioeconomic status act as critical determinants of health, with vulnerable populations facing greater structural barriers to accessing diagnostic examinations and specialized oncology services, resulting in unfavorable clinical outcomes and lower survival rates.[13] Thus, the rise of EOCRC represents not only an individual clinical challenge but also a public health imperative that requires reassessment of awareness strategies and optimization of screening criteria for young adults.[7]

One of the main obstacles to the timely diagnosis of early-onset colorectal cancer (EOCRC) is the low level of clinical suspicion when patients present with the initial manifestations of the disease.[9] Symptoms such as hematochezia, abdominal pain, changes in bowel habits, and iron-deficiency anemia are frequently attributed to benign conditions prevalent among younger individuals, such as hemorrhoidal disease, irritable bowel syndrome, or inflammatory bowel diseases.[3] Although hematochezia is one of the earliest and most prevalent warning signs, it is repeatedly underestimated by both patients and healthcare professionals due to the misconception that oncological risk is low in this age group. This delays essential endoscopic investigations and allows silent tumor progression.[2]

The nonspecific nature of symptoms such as fatigue and intermittent abdominal discomfort further contributes to diagnostic difficulties, as patients often undergo multiple medical consultations and successive ineffective treatments before the tumor is confirmed.[3] In several cases, EOCRC is identified only after investigation of advanced cases of persistent anemia or unexplained weight loss, highlighting the failure to recognize early warning signs that appear relatively nonspecific.[14]

This situation is aggravated by the absence of systematic screening among asymptomatic young individuals. Historically, the exclusion of individuals younger than 50 years from screening protocols created a gap in clinical surveillance, leaving younger populations without preventive care in the absence of a family history that would justify early screening.[8] Although recent recommendations have lowered the starting age for these examinations, a large proportion of individuals younger than this new age threshold remain without preventive follow-up.[4]

Consequently, the interval between symptom onset and definitive diagnosis tends to be significantly longer than that observed in older adults, resulting in disease detection predominantly at locally advanced or metastatic stages.[2,3] This delayed diagnosis necessitates more aggressive therapeutic interventions, increasing morbidity and drastically reducing overall survival rates.[6]

The prolonged diagnostic interval contributes to the predominant clinical presentation at advanced stages (III and IV), severely compromising prognosis.[3] This situation is further aggravated by the fact that early-onset colorectal cancer (EOCRC) frequently exhibits more aggressive biological behavior. Histologically, there is a higher incidence of poorly differentiated neoplasms, mucinous

tumors, and signet-ring cell carcinomas.[10] These characteristics confer greater invasive potential, extensive lymph node involvement, and an increased risk of metastatic spread at the time of detection.[11] Furthermore, the literature indicates that EOCRC has a molecular profile distinct from that observed in older patients, reinforcing the hypothesis that specific pathophysiological pathways drive this aggressiveness in younger individuals.[15]

From a clinical perspective, late identification of the disease results in increased morbidity and mortality. Young patients diagnosed at advanced stages have a greater need for prolonged hospitalizations, emergency surgical interventions, and palliative therapies, substantially increasing healthcare costs and reducing the likelihood of treatment with curative intent.[9] Although young adults demonstrate better physiological tolerance to antineoplastic therapies due to the lower prevalence of preexisting comorbidities, this potential advantage is frequently offset by the tumor burden and metastatic spread present at diagnosis.[2]

Beyond the strictly physiological consequences, the psychosocial burden associated with EOCRC is disproportionately high. Unlike older populations, individuals diagnosed at a young age are at the peak of their productive years. The need for intensive multimodal treatments—including chemotherapy, radiotherapy, and extensive surgical resections—has profound and long-term effects on fertility, sexual health, professional stability, and mental health.[16] The abrupt disruption of daily routines and life plans causes severe physical and emotional impairment, requiring a multidisciplinary approach capable of addressing the specific needs of this age group and extending beyond the mere assessment of overall survival.[8]

The molecular architecture of early-onset colorectal cancer (EOCRC) reveals a significant contribution from genetic factors, particularly in cases diagnosed at very young ages or in patients with a family history of cancer.[1] In this context, hereditary syndromes such as Lynch syndrome and familial adenomatous polyposis (FAP) are among the main predisposing factors, increasing the lifetime risk of developing malignant lesions.[5] DNA mismatch repair deficiency (dMMR) is frequently reported among young patients with colorectal cancer and is closely associated with microsatellite instability.[15] Despite the importance of this testing for diagnosis and treatment selection, low adherence to molecular screening recommendations among young adults is observed in clinical practice, representing a significant failure in identifying patients at high hereditary risk.[5]

It is noteworthy that many young patients with EOCRC have no known family history, suggesting that sporadic mutations and environmental factors also exert a strong influence on the development of these early-onset tumors.[8] Therefore, the absence of a family history should not be used to rule out clinical suspicion or limit genetic investigation in this population.[4] This biological profile is often accompanied by aggressive histological characteristics, such as the presence of signet-ring cells and mucinous histology, patterns that favor more rapid disease progression and hinder detection at early stages.[9]

Furthermore, the lack of adequate molecular testing, often due to resource shortages or shortcomings in hospital protocols, compromises not only patient care but also the protection of family members.[15] When a hereditary syndrome is not identified in a timely manner, the opportunity to perform preventive examinations in first-degree relatives is lost, reducing the chances of surveillance and early diagnosis among these individuals.[4] Thus, expanding access to genetic and molecular testing among young adults becomes a necessary tool for understanding individual risk and implementing more effective prevention strategies for patients and their families.[1]

Social determinants of health, such as ethnicity, socioeconomic status, and educational attainment, exert a decisive influence on early diagnosis and clinical outcomes in early-onset colorectal cancer (EOCRC).[13] Data analysis demonstrates that these variables, together with the quality of healthcare

coverage, are directly associated with diagnosis at advanced stages and lower overall survival rates.[9] Patients experiencing social vulnerability or having limited access to health insurance face structural barriers that hinder access to specialist consultations and complex diagnostic procedures, such as colonoscopy.[3] In these cases, persistent symptoms without appropriate investigation over prolonged periods favor silent tumor progression and compromise the final prognosis.[13]

Racial disparities also emerge as a critical factor, with certain ethnic groups recording higher incidence and mortality rates at younger ages.[17] This phenomenon does not merely reflect biological differences but results from a combination of inequalities in access to timely diagnosis and structural deficiencies in the support provided by healthcare systems.[15] The lack of adequate health insurance coverage prolongs the interval between the initial complaint and confirmation of the tumor, which explains the higher frequency of metastatic disease among socioeconomically disadvantaged groups.[13] Conversely, individuals with easier access to screening services and specialized follow-up tend to experience better oncological outcomes.[7]

In addition to financial and structural obstacles, cultural barriers and low awareness of EOCRC contribute to delays in seeking medical care. The misconception that colorectal cancer affects only older individuals leads both patients and healthcare professionals to minimize symptoms, with clinicians often maintaining a low level of diagnostic suspicion when evaluating young adults.[2] This situation reinforces the need for strategies that extend beyond technical advances, focusing on reducing inequalities in healthcare access, strengthening screening policies, and implementing health education campaigns to raise awareness among the population and primary care providers.[9]

Traditional colorectal cancer (CRC) screening strategies, historically targeted at individuals older than 50 years, have demonstrated critical limitations in the face of increasing early-onset colorectal cancer (EOCRC) incidence.[7] Although recent international guidelines have lowered the starting age for screening average-risk individuals to 45 years, available data indicate that a substantial proportion of young patients are diagnosed at ages below this new threshold.[9] This situation suggests that simply lowering the chronological age threshold, although necessary, is insufficient as a standalone measure to prevent delayed diagnosis. Reliance on fixed protocols ultimately overlooks symptomatic individuals who remain outside the scope of conventional screening strategies.[3]

Another relevant aspect concerns healthcare professionals' awareness of colorectal cancer in young adults. The tendency to underestimate warning signs in this age group delays the ordering of diagnostic examinations, reinforcing the need for medical education that prioritizes clinical suspicion regardless of patient age.[2] At the same time, public awareness campaigns are essential to challenge the misconception that the disease affects only older individuals, encouraging prompt medical evaluation when symptoms arise that patients themselves often ignore or minimize.[14]

The transition toward a more effective surveillance model requires individualized screening strategies that consider family history, genetic predisposition, and the presence of associated risk factors.[1] Patients with hereditary syndromes, a family history of the disease, or suspicious clinical manifestations should undergo closer follow-up and earlier initiation of colonoscopic surveillance.[5] Establishing an integrated healthcare network that combines a high level of diagnostic suspicion, facilitated access to imaging examinations, and genetic support is essential to reverse the trend toward advanced-stage diagnosis among younger populations, improving prognosis and reducing the social impact of this malignancy during the productive years of life.[9]

CONCLUSION

The findings demonstrate that diagnostic delays in colorectal cancer among young adults are multifactorial, primarily resulting from low clinical suspicion in the presence of nonspecific initial symptoms—frequently attributed to benign conditions—and the historical exclusion of this age group from systematic screening protocols. Consequently, early-onset colorectal cancer is predominantly diagnosed at locally advanced or metastatic stages. This situation necessitates more invasive therapeutic approaches and has severe psychosocial consequences, increasing morbidity and mortality among individuals at the peak of their productive years.

Although this study has identified these challenges, it is important to acknowledge certain limitations, including the nature of the integrative review, which brings together studies with different methodologies and sociodemographic contexts, as well as the scarcity of epidemiological data focusing exclusively on individuals younger than 45 years, which may limit the unrestricted generalizability of the findings.

In this regard, further research should include prospective multicenter studies focused on the genetic and molecular characterization of tumors in this age group, as well as evaluations of the impact of awareness campaigns and continuing medical education on reducing time to diagnosis. It is also recommended that the feasibility of personalized screening models based on individual risk stratification, family history, and social determinants of health be investigated.

In summary, addressing colorectal cancer in young adults goes beyond simply lowering screening age thresholds, requiring an urgent paradigm shift in clinical suspicion and the strengthening of public policies that ensure preventive, accessible, and equitable oncological care.

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