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OPINION ARTICLE

Colorectal cancer: towards a new era of early detection and personalized medicine

Cáncer colorrectal: hacia una nueva era de detección precoz y medicina personalizada

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ABSTRACT

Colorectal cancer is the second leading cause of cancer death worldwide and represents a significant burden on healthcare systems. This opinion piece analyzes the evolution of early detection strategies, from extending the screening age to 45 years to incorporating liquid biomarkers and advanced molecular techniques. Changes in international guidelines, the role of genetics in risk stratification, and the challenges of implementing effective population-based programs are discussed. The article concludes that optimizing colorectal

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cancer screening requires a multidimensional approach that integrates technological innovation, health education, and equitable access.

Keywords: colorectal cancer, screening, early detection, liquid biomarkers, Lynch syndrome, colonoscopy

RESUMEN

El cáncer colorrectal representa la segunda causa de muerte por cáncer a nivel mundial y una carga sanitario significativa para los sistemas de salud. El presente artículo de opinión analiza la evolución de las estrategias de detección precoz, desde la ampliación de la edad de inicio del tamizaje hasta los 45 años hasta la incorporación de biomarcadores líquidos y técnicas moleculares avanzadas. Se discuten los cambios en las guías internacionales, el papel de la genética en la estratificación del riesgo y los desafíos para la implementación de programas poblacionales efectivos. Se concluye que la optimización del cribado colorectal requiere un enfoque multidimensional que integre innovación tecnológica, educación sanitaria y equidad en el acceso.

Palabras clave: cáncer colorrectal, tamizaje, detección precoz, biomarcadores líquidos, síndrome de Lynch, colonoscopía

Colorectal cancer (CRC) is the most common neoplasm of the digestive system and the second leading cause of global cancer mortality [1]. Despite its high prevalence and the availability of effective detection methods, a significant proportion of cases are diagnosed at advanced stages, limiting therapeutic options and compromising survival [2]. The World Health Organization has identified CRC as a priority in its cancer control strategies, emphasizing early detection as a fundamental pillar for reducing the disease burden [9].

In recent years, the colorectal screening paradigm has undergone substantial transformations driven by epidemiological evidence and technological advances [3-8]. Lowering the screening initiation age to 45, the incorporation of blood tests based on circulating tumor DNA, and the personalization of strategies according to genetic profile represent advances that redefine the preventive approach to this pathology [12, 15]. The objective of this article is to analyze these innovations and their implications for clinical practice and public health.

Expanding Screening: The Evidence that Changed the Guidelines

The decision to expand colorectal screening to adults aged 45 to 49, adopted by the U.S. Preventive Services Task Force (USPSTF) in 2021 and endorsed by the American Cancer Society (ACS) and the National Comprehensive Cancer Network (NCCN), responded to the documented increase in CRC incidence in the younger population [12, 16]. This modification, which elevated the recommendation to grade B for the 45-49 age group, has demonstrated a positive impact on the detection of precursor lesions [16].

Recent studies using electronic health records have confirmed that following the implementation of this guideline, a substantial increase in the performance of colonoscopies with pathological findings was observed in the 45-49 age group, with higher detection of precancerous polyps and lower identification of invasive cancers compared to older age groups [14]. The authors consider that these findings suggest the expansion of screening is achieving its primary objective: the identification and resection of lesions before their progression to malignancy.

However, the authors suggest that the universal implementation of this recommendation faces logistical and resource challenges, particularly in healthcare systems with limited coverage and disparities in access to endoscopy [11]. Colonoscopy, although considered the gold standard, requires specialized infrastructure and trained personnel, elements not always available in all healthcare contexts [11].

Liquid Biopsies: The Future of Non-Invasive Screening

The development of blood tests for colorectal cancer detection represents one of the most significant advances in the field of screening. In 2025, the NCCN incorporated the Shield test, an assay based on circulating tumor DNA (ctDNA), into its guidelines, recommending its performance every three years in the average-risk population [15].

The ECLIPSE study, published in the New England Journal of Medicine, demonstrated that this test has 83% sensitivity for detecting CRC, with the advantage of being non-invasive and easily accessible during any medical consultation [15]. The authors consider that the inclusion of such technologies in clinical practice guidelines marks a turning point towards massive population screening, particularly in populations reluctant to undergo endoscopic procedures or with geographical limitations in accessing colonoscopy.

However, the authors note that ctDNA tests must be interpreted with caution: a positive result requires colonoscopic confirmation, and their sensitivity for precursor lesions (advanced adenomas) is inferior to direct colonoscopy [15]. Therefore, these tests do not replace but rather complement existing strategies, offering an option to increase screening adherence in individuals who would otherwise not undergo screening [15].

Genetic Stratification: Beyond Average Risk

The recognition of hereditary syndromes such as Lynch syndrome (LS) has transformed the approach to CRC in the high-risk population. Updated guidelines from the Chinese Society of Clinical Oncology (CSCO) 2025 and the NCCN emphasize the importance of systematic genetic evaluation and intensive surveillance in mutation carriers [10, 12].

For individuals with LS, current recommendations indicate starting annual or biennial colonoscopy from age 25, given the aggressive behavior of these tumors and their predilection for the proximal colon [10, 13]. The authors propose that the identification of mutations in DNA repair genes (MLH1, MSH2, MSH6, PMS2) and the implementation of hereditary tumor registries enable effective secondary prevention, with the potential to reduce mortality in these families [10].

Furthermore, the detection of POLE/POLD1 mutations has emerged as a predictive factor for response to immune checkpoint inhibitors, opening new therapeutic perspectives even in tumors with low or stable microsatellite instability. The authors consider that precision medicine in CRC transcends the therapeutic realm to integrate into personalized prevention and surveillance strategies [10].

Post-Surgical Surveillance: The Role of Circulating DNA

The monitoring of resectable CRC has evolved with the introduction of circulating tumor DNA analysis to detect minimal residual disease (MRD). The CSCO 2024-2025 guidelines include ctDNA assessment as a class III recommendation for postoperative surveillance in stages I-III, recognizing its potential to predict recurrence and guide adjuvant chemotherapy decisions [10].

The authors suggest that, although the standardization of these techniques is still under development, tumor-informed assays (personalized according to the mutational profile of the primary tumor) offer greater sensitivity and

specificity than fixed panels. This approach allows for the detection of recurrence months before radiological evidence, potentially improving outcomes through early intervention [10].

Challenges in Implementing Screening Programs

Despite the scientific robustness of colorectal screening, population participation remains suboptimal. In Spain, although organized programs exist in several autonomous communities, adherence rates are lower than those of other cancer screening programs, attributable in part to the perception of invasiveness of colonoscopy and lack of awareness about the importance of prevention [11].

The authors suggest that diversifying screening options —including annual fecal immunochemical testing, fecal DNA tests every 1-3 years, and now liquid biopsies— can improve population coverage [11]. However, they emphasize that any strategy must guarantee the quality of follow-up for positive results, preventing the expansion of first-level options from compromising the effectiveness of the overall program.

Colorectal cancer exemplifies how the convergence of epidemiology, genomics, and diagnostic technology can transform cancer prevention [1, 10]. Recent advances —expanding screening to age 45, incorporating ctDNA tests, genetic stratification, and post-surgical molecular surveillance— offer unprecedented tools to reduce the burden of this disease [10, 12, 15].

The authors conclude that the success of these strategies depends on overcoming implementation barriers, ensuring equity in access, and maintaining rigor in the quality of programs [11]. Health education, professional training, and investment in genomic and endoscopic infrastructure are indispensable pillars for realizing the potential of these innovations. Finally, the authors propose that we, as coloproctologists, must lead the integration of these technologies into coherent clinical workflows, promoting a care model that combines molecular precision with sensitivity to each patient's needs and preferences.

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CONFLICT OF INTEREST

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