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# Comparative Study on the Application Effect of Molecular Detection in the Early Diagnosis of Colorectal Cancer

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**Abstract:** *Objective*: To comparatively analyze the application value of molecular detection and traditional screening methods in the early diagnosis of colorectal cancer (CRC), clarify the detection efficacy of molecular detection for early CRC and precancerous lesions, and provide evidence-based support for optimizing early diagnosis strategies in clinical practice. *Methods*: A total of 200 patients with suspected colorectal lesions admitted to our hospital from January 2025 to January 2026 were selected as the study subjects. Using colonoscopy combined with histopathological examination as the diagnostic gold standard, patients were randomly assigned to an observation group (100 cases) and a control group (100 cases) using a random number table method. The observation group underwent fluorescence in situ hybridization (FISH) molecular detection, while the control group received a traditional screening protocol combining serum tumor markers with fecal immunochemical testing (FIT). Diagnostic sensitivity, specificity, positive predictive value (PPV), and negative predictive value (NPV) were compared between the two groups. The detection rates of CRC at different stages and precancerous adenomas were analyzed, and screening compliance and missed diagnosis rates were recorded for both groups. *Results*: Based on pathological results, among the 200 patients, 56 were diagnosed with CRC (33 with stage I-II early-stage cancer and 23 with stage III-IV advanced cancer), 20 with high-grade intraepithelial neoplasia, 41 with low-grade adenomas, and 83 with benign lesions or normal findings. The observation group demonstrated significantly higher diagnostic sensitivity, specificity, PPV, and NPV for CRC compared to the control group (all $P < 0.05$). The detection rates of early-stage CRC and precancerous adenomas were significantly higher in the observation group than in the control group. Both groups showed high detection rates for advanced CRC, with no significant difference between them ($P > 0.05$). Screening compliance was significantly higher, and the missed diagnosis rate was significantly lower in the observation group than in the control group (both $P < 0.05$). *Conclusion*: FISH molecular detection exhibits superior sensitivity and specificity in the early diagnosis of CRC, with distinct advantages in detecting early-stage cancer and precancerous lesions. It also offers better patient compliance and a lower missed diagnosis rate, making it a preferred option for early screening in high-risk populations for CRC. This approach can complement traditional screening methods and facilitate early diagnosis and treatment of CRC.

**Keywords:** Colorectal cancer; Early diagnosis; Molecular detection; Circulating tumor DNA; Gene methylation; Comparative study

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# 1. Introduction

Colorectal cancer (CRC) is a highly prevalent malignant gastrointestinal tumor worldwide. According to data from the National Cancer Center, CRC ranks among the leading causes of cancer incidence and mortality among gastrointestinal tumors in China, with a rising trend toward younger onset. Individuals aged 45 and above constitute the high-risk group, and the incidence is significantly higher in urban areas than in rural areas [1]. Prognosis is directly correlated with clinical stage; the 5-year survival rate exceeds 90% for patients with early-stage disease following standardized treatment but drops below 30% for those with advanced disease. Most patients with advanced CRC are diagnosed after missing the optimal window for surgical intervention. Therefore, standardized early screening is crucial for reducing mortality and improving prognosis [2]. Colonoscopy combined with pathological biopsy remains the gold standard for CRC diagnosis. However, as an invasive procedure requiring bowel preparation and often causing discomfort, it is associated with low public compliance and is unsuitable for widespread population-based screening [3]. Recent advances in molecular biology have facilitated the application of fluorescence in situ hybridization (FISH) technology in early CRC diagnosis. FISH offers advantages such as intuitive localization, high specificity, rapid detection, and compatibility with various sample types, enabling precise identification of genetic abnormalities and addressing limitations of traditional screening methods. This study conducted a prospective comparative analysis to evaluate the early diagnostic efficacy of two screening protocols and assess the detection performance of molecular testing, providing practical evidence for establishing an efficient early diagnosis system.

# 2. Materials and methods

## 2.1. General information

A total of 200 patients with suspected colorectal lesions admitted to the Department of Gastroenterology and Oncology at our hospital from January 2023 to January 2025 were selected as the study subjects. Using a random number table method, patients were divided into an observation group and a control group, with 100 cases in each group. Inclusion criteria were as follows: (1) aged 40–75 years old with risk factors for CRC; (2) presence of gastrointestinal symptoms such as hematochezia, mucous stool, abdominal pain, or incomplete evacuation, or abnormal intestinal findings on routine physical examination; (3) voluntary participation in the study, provision of informed consent, and cooperation in completing all screening procedures and colonoscopy; (4) no history of colorectal surgery, intestinal malignancy, severe liver or kidney dysfunction, or coagulopathy. Exclusion criteria were as follows: (1) concurrent malignancy at other sites; (2) recent receipt of antitumor therapy such as radiotherapy, chemotherapy, or targeted therapy; (3) inability to tolerate colonoscopy or molecular testing; (4) incomplete clinical data.

The observation group comprised 54 males and 46 females, aged 41-74 years (mean age, 58.62 ± 7.35 years old). Risk factor distribution included a family history of CRC in 20 cases, chronic colitis in 18 cases, a history of intestinal polyps in 26 cases, and abnormal bowel habits in 36 cases. The control group comprised 53 males and 47 females, aged 40–75 years old (mean age, 57.98 ± 7.61 years old). Risk factor distribution included a family history of CRC in 19 cases, chronic colitis in 20 cases, a history of intestinal polyps in 27 cases, and abnormal bowel habits in 34 cases. No significant differences in gender, age, or risk factor distribution were observed between the two groups ($P > 0.05$), indicating comparability.

## 2.2. Methods

### 2.2.1. Control group

The control group underwent screening combining serum tumor markers with FIT. (1) Serum tumor marker detection: Five milliliters of fasting venous blood were collected from each patient, and serum was separated by centrifugation. Levels of carcinoembryonic antigen (CEA), carbohydrate antigen 19-9 (CA19-9), and carbohydrate antigen 72-4 (CA72-4) were measured using chemiluminescent immunoassay on an automated chemiluminescent immunoanalyzer, following the reagent kit instructions. Positive results were defined as CEA > 5.0 ng/mL, CA19-9 > 37.0 U/mL, or CA72-4 > 6.9 U/mL. (2) FIT: Fresh stool samples were collected from patients, and fecal occult blood was detected using an immunocolloidal gold method. A positive result was indicated by the presence of a positive band. A positive result on either test was considered a positive traditional screening result, necessitating further colonoscopy.

### 2.2.2. Observation group

The observation group underwent FISH to detect colorectal exfoliated cells or lesional tissue, with strict adherence to cytogenetic protocols. (1) Sample collection: For patients who had not undergone colonoscopy, fecal samples or intestinal mucosal exfoliated cells obtained via endoscopic brushing were collected. For patients who had undergone preliminary endoscopic examination, active tissue samples were obtained from lesional sites, avoiding necrotic areas. (2) Sample processing: Cell samples were fixed, mounted on slides, dehydrated, and digested with protease to remove impurities. Tissue samples were embedded in paraffin, sectioned, deparaffinized, and hydrated before preprocessing. (3) FISH hybridization detection: Specific fluorescently labeled probes targeting CEP8, c-Myc, and DCC were applied to slides, which were then sealed, denatured, and hybridized overnight at a constant temperature. Result interpretation: After hybridization, free probes were washed away, and cell nuclei were counterstained with DAPI. Under a 100× oil immersion lens, 100 intact cell nuclei were counted. The presence of amplified, deleted, or abnormally located fluorescent signals was considered a positive result. Positive cases required further colonoscopy and pathological biopsy for confirmation.

### 2.2.3. Gold standard diagnosis

All patients underwent electronic colonoscopy, and multiple biopsies were taken from intestinal lesions for histopathological examination. Pathological diagnosis, considered the gold standard, classified patients into CRC (stage I-II early-stage cancer, stage III-IV advanced cancer), high-grade intraepithelial neoplasia, low-grade adenoma, or benign lesions/normal. Two senior pathologists independently interpreted the results, with discrepancies resolved through consultation to determine the final diagnosis.

## 2.3. Observation indicators

(1) Diagnostic efficacy indicators: Using pathological results as the gold standard, sensitivity, specificity, PPV, and NPV were calculated for both screening protocols. Sensitivity = (true positives)/(true positives + false negatives) × 100%; specificity = (true negatives)/(true negatives + false positives) × 100%; PPV = (true positives)/(true positives + false positives) × 100%; NPV = (true negatives)/(true negatives + false negatives) × 100%.

(2) Lesion stratification detection: Detection rates of early-stage CRC (stage I-II), advanced CRC (stage III-IV), and precancerous adenomas were compared between the two groups.

(3) Compliance and missed diagnosis: Screening compliance rate (number of patients completing all screening procedures/total number of patients × 100%) and missed diagnosis rate (number of false negatives/total number of confirmed lesions × 100%) were calculated for both groups.

### 2.4. Statistical methods

Data analysis was performed using SPSS 25.0 statistical software. Continuous data are presented as mean ± standard deviation (SD), with intergroup comparisons conducted using the t-test. Categorical data are presented as [n (%)], with intergroup comparisons conducted using the $\chi^2$ test. A $P$-value < 0.05 was considered statistically significant.

## 3. Results

### 3.1. Pathological diagnosis results

Out of 200 patients, diagnosis was confirmed through colonoscopy combined with pathological biopsy: 56 cases of colorectal cancer were identified, including 33 cases of early-stage cancer (Stage I-II) and 23 cases of middle to advanced-stage cancer (Stage III-IV); 20 cases of high-grade intraepithelial neoplasia; 41 cases of low-grade adenomas; 30 cases of benign intestinal conditions such as inflammation and polyps; and 53 cases from the normal population.

### 3.2. Comparison of diagnostic efficacy between the two groups

The sensitivity, specificity, positive predictive value, and negative predictive value for colorectal cancer diagnosis in the observation group were significantly higher than those in the control group (all $P < 0.05$). See **Table 1** for details.

**Table 1.** Comparison of diagnostic efficacy for colorectal cancer between the two groups [n/%]

| Group | Sensitivity (%) | Specificity (%) | Positive Predictive Value (%) | Negative Predictive Value (%) |
|---|---|---|---|---|
| Observation Group (n = 100) | 87.50 (49/56) | 90.48 (76/84) | 84.00 (49/58) | 92.68 (76/82) |
| Control Group (n = 100) | 62.50 (35/56) | 73.81 (62/84) | 67.57 (35/52) | 69.44 (62/89) |
| $\chi^2$ | 9.333 | 7.954 | 4.481 | 14.522 |
| $P$ | 0.002 | 0.005 | 0.034 | 0.000 |

### 3.3. Comparison of detection rates for different lesion stratifications between the two groups

The detection rates of early-stage colorectal cancer and precancerous adenomas in the observation group were significantly higher than those in the control group. Both groups exhibited relatively high detection rates for advanced colorectal cancer, with no statistically significant difference observed between them ($P > 0.05$). See **Table 2** for details.

**Table 2.** Comparison of detection rates for different lesion stratifications between the two groups

| Lesion Type | Total Cases | Detection Cases in Observation Group (%) | Detection Cases in Control Group (%) | $\chi^2$ | *P* |
|---|---|---|---|---|---|
| Early-stage colorectal cancer (Stage I-II) | 33 | 28 (84.85) | 16 (48.48) | 9.818 | 0.002 |
| Advanced colorectal cancer (Stage III-IV) | 23 | 21 (91.30) | 19 (82.61) | 0.192 | 0.662 |
| Premalignant adenoma | 41 | 24 (58.54) | 11 (26.83) | 8.424 | 0.004 |

## 3.4. Comparison of screening compliance and missed diagnosis rates

Between the Two Groups The screening compliance rate in the observation group was significantly higher than that in the control group, while the missed diagnosis rate was significantly lower (both $P < 0.05$). See **Table 3** for details.

**Table 3.** Comparison of screening compliance and missed diagnosis rates between the two groups

| Group | Compliance Rate [n (%)] | Missed Diagnosis Rate [n (%)] |
|---|---|---|
| Observation Group (n = 100) | 91 (91.00) | 7 (12.50) |
| Control Group (n = 100) | 76 (76.00) | 21 (37.50) |
| $\chi^2$ | 8.165 | 7.236 |
| *P* | 0.004 | 0.007 |

# 4. Discussion

The development of colorectal cancer is a multigene, multistage evolutionary process. It typically takes 5 to 10 years for the progression from normal mucosa to adenoma and then to carcinogenesis, providing a sufficient window for early screening [4]. The core objective of clinical screening is to achieve precise identification at the precancerous or early cancerous stage and to block disease progression through timely intervention [5]. In traditional screening protocols, serum tumor markers are commonly used as non-invasive screening methods in clinical practice. However, due to the extremely low levels of markers released by tumor cells in early-stage carcinogenesis, the detection sensitivity is insufficient, and they are easily interfered with by factors such as inflammation and benign tumors, resulting in a high false-positive rate [6]. FIT (fecal immunochemical test) can only identify hemorrhagic lesions in the intestine and has poor detection capabilities for early-stage cancers without bleeding and flat adenomas, leading to a high risk of missed diagnoses. In this study, the overall sensitivity of the traditional screening protocol in the control group for colorectal cancer was only 62.50%, with an early cancer detection rate of less than 50% and a precancerous adenoma detection rate of only 26.83%, fully confirming the limitations of traditional methods in early diagnosis.

Fluorescence in situ hybridization (FISH) is a precise molecular cytogenetic technique based on the principle of nucleic acid base complementary pairing. It can directly localize target genes at the cellular or tissue level, visually identifying specific genetic alterations in colorectal cancer, such as gene amplification, deletion, and copy number abnormalities. It is not affected by factors such as tumor bleeding, clinical stage, or protein expression fluctuations, enabling the capture of abnormal changes at the cellular genetic level in the early stages of lesions

and significantly improving the detection efficiency of early lesions [7]. Compared with other molecular detection techniques, FISH does not require DNA extraction or PCR amplification, avoiding result deviations caused by nucleic acid degradation and amplification contamination. The detection results are intuitive, with extremely high specificity. Moreover, it can be adapted to both colorectal exfoliated cells and tissue samples, making it suitable for both preoperative non-invasive screening and intraoperative rapid pathological auxiliary diagnosis, with broader clinical applicability. In the observation group of this study, a combination of probes targeting the CEP8 chromosome, c-Myc gene, and DCC gene was selected, covering core genetic abnormality targets in the development of colorectal cancer. The combined interpretation of multiple targets further reduced the false-positive and false-negative rates, significantly improving diagnostic accuracy.

The results of this study showed that the diagnostic sensitivity and specificity of the molecular detection protocol in the observation group both exceeded 85%, significantly outperforming the traditional screening protocol. In particular, the detection sensitivity for stage I-II early-stage colorectal cancer reached 84.85%, nearly doubling that of the control group, and the detection rate for precancerous adenomas also increased to 58.54%, fully demonstrating the core advantages of molecular detection in early lesion identification. The main reason for this is that molecular detection targets tumor-specific molecular markers rather than indirect bleeding or protein expression abnormalities, resulting in a lower false-positive rate and higher specificity [8]. Meanwhile, molecular detection is non-invasive, with fecal samples that can be collected at home and blood samples that are easy to obtain, without the need for bowel preparation or invasive procedures, leading to higher patient compliance. In this study, the compliance rate in the observation group reached 91.00%, much higher than that in the control group. High compliance can effectively improve screening coverage and reduce missed diagnoses due to refusal of examination.

In terms of the missed diagnosis rate, the observation group had a missed diagnosis rate of only 12.50%, while the control group had a high rate of 37.50%. The high missed diagnosis rate of traditional screening protocols is mainly due to factors such as the lack of obvious molecular marker expression in early lesions and the absence of bleeding in small adenomas. Molecular detection, however, can detect genetic abnormalities when lesions have not yet exhibited obvious morphological changes or clinical symptoms, achieving “early detection and early diagnosis.” In addition, the detection rate of molecular detection for middle-to-late-stage colorectal cancer is similar to that of traditional protocols, indicating that it does not miss advanced lesions and meets the screening needs for both early and middle-to-late-stage lesions, with broader applicability.

Fluorescence in situ hybridization (FISH) molecular detection technology has significant clinical advantages in the early diagnosis of colorectal cancer [9,10]. Compared with the traditional screening protocol combining serum tumor markers and FIT, it has higher diagnostic sensitivity and specificity, outstanding capabilities in identifying cytogenetic abnormalities in early-stage colorectal cancer and precancerous adenomas, intuitive and reliable detection results, better patient screening compliance, and a lower missed diagnosis rate. It can effectively compensate for the technical deficiencies of traditional screening methods. Incorporating FISH technology into the early screening system for colorectal cancer and prioritizing non-invasive exfoliated cell FISH screening for high-risk populations can achieve precise early diagnosis of colorectal cancer, provide a reliable basis for clinical early intervention, thereby reducing the mortality rate of colorectal cancer and improving the overall prognosis of patients, with significant clinical promotion value. Subsequent studies can further expand the sample size, conduct multicenter, long-term follow-up studies, optimize FISH probe combinations, simplify operational procedures, and promote its widespread application in primary healthcare institutions.

## 5. Conclusion

FISH molecular detection is a highly sensitive and specific tool for early CRC diagnosis, offering key advantages in patient compliance and reduced misdiagnosis. It serves as an effective complement to conventional screening methods, supporting earlier intervention in high-risk populations.


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## Disclosure statement

The authors declare no conflict of interest.

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