THE VALUE OF FAMILY HISTORY IN CANCER RISK ASSESSMENT

Family history is a powerful screening tool.

In conjunction with the patient's medical history, family history can inform diagnosis, promote risk assessment, and prevent, detect and manage disease. This is especially true for cancer.

When it works

Family history is most useful when it is available in a structured format in the medical record and of course, when it is accurate and complete to support risk assessment. Not all family history information is equal. Seeing that a patient has a "family history of cancer" in the medical record is not specific enough to allow for immediate analysis; seeing documentation that the patient's mother had colon cancer at age 53 allows for personalized risk assessment and possibly, a change in screening regimen.

How it works

The goal of family history risk assessment is to identify individuals with strong and moderate genetic predispositions to disease so that they can adopt prevention or screening activities to reduce risk and detect disease early. The risk assessment process starts by identifying red flags and patterns in the patient's family history, and then uses that information to stratify individuals into average, increased, or high risk.

Necessary for guidelines-based screening

National guidelines recommend earlier and more frequent screening for individuals at increased risk for CRC. For individuals at high (hereditary) risk, additional evaluations and health services may be indicated, such as genetic testing or prophylactic surgery. In order to accurately identify the best cancer management plan for each patient, clinicians must assess the family history.

Extra benefits

In addition to its critical role in risk assessment, the act of family history collection can be a benefit to the patient, as can the discussion about the family history between patient and provider. The process of eliciting a family history provides an excellent opportunity to build a relationship with the patient and to become aware of the patient's motivations and concerns. Such information can be beneficial as the provider helps the patient make health-related decisions. The emphasis on disease prevention and management based on the family history may motivate changes in behavior that forestall disease or reduce its adverse effects.

Eliciting and summarizing family history information can:

- help the patient understand the condition in question,
- clarify patient misconceptions,
- demonstrate variation in disease expression (such as different ages at onset),
- provide a reminder of who in the family is at risk for the condition, and
- emphasize the need to obtain medical documentation on affected relatives.

See best practices in family history collection and risk assessment for primary care in the Appendix.

THE IMPORTANCE OF IDENTIFYING COLORECTAL CANCER FAMILY HISTORY

Colorectal cancer can be prevented when we know who is at increased risk.

Colorectal cancer (CRC) is the second leading cause of cancer deaths in the United States. In 2018, there are predicted to be 140,250 new cases of CRC in the United States.[2] Individuals who have a first-degree relative with CRC are at least two times more likely to develop CRC themselves, with the risk increasing with earlier ages of diagnosis and the number of relatives diagnosed with CRC.[3,4] Therefore, knowledge of and adherence to screening guidelines is important to improve morbidity and mortality from CRC in these families at increased risk.

Routine screening has been shown to be effective in prevention and early detection of CRC. Early detection of CRC saves lives. The survival rate for patients with stage 1 (local) CRC is 90% but drops to 14% for patients with stage 4 (metastatic) disease.[2] Approximately 4,600 lives could be saved per year if individuals with CRC under age 50 are diagnosed at a localized stage.

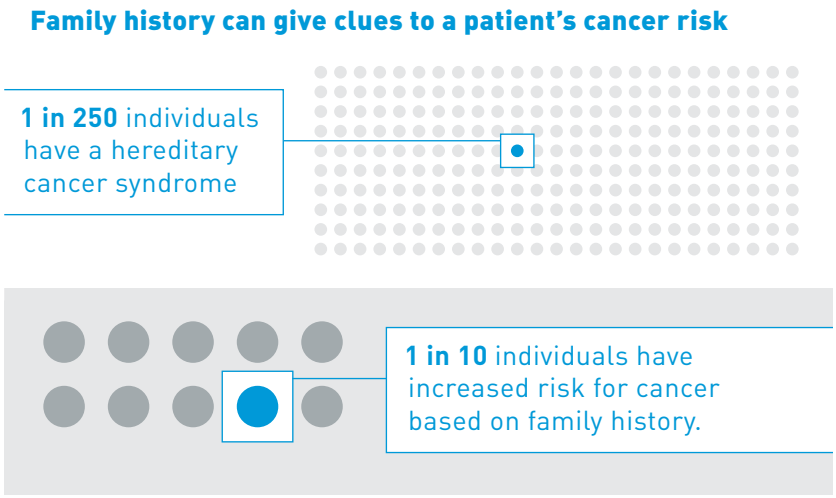

Figure 1. Incidence of familial and hereditary cancer risks for colon and breast cancers.

National screening guidelines exist for the general population at average risk, for individuals at moderately increased risk due to a positive family history and/or personal history, and for those at high risk due to a hereditary cancer syndrome. However, fewer than half of individuals with a family history of CRC or advanced adenoma (> 1 cm) receive personalized counseling and follow risk-based screening guidelines.[4,5]

This concerning state is due in part to a lack of family history collection among a significant number of patients. Less than 40% of individuals with a family history of CRC have talked with a healthcare provider about their family history.[5] Even in symptomatic patients with rectal bleeding, family history is not always adequately collected, with 38% of cases lacking necessary information for risk evaluation.[6] Expanding beyond CRC to include additional common conditions in primary care, one study showed that less than 4% of patients' medical records had sufficient family history information to assess risk.[7]

Limited or inaccurate family history collection and risk assessment is a major barrier to successful cancer screening. In order to focus screening and prevention efforts on those with familial or hereditary risk, these individuals must first be identified as having an increased risk, which requires collecting the necessary family history information for risk assessment. Primary care clinicians play a pivotal role in identifying people at increased CRC risk and facilitating recommended screening. This toolkit aims to help the clinician implement best practices in CRC family history collection, risk assessment, and management to prevent cancer or detect it at the earliest possible stage.

EARLY ONSET COLORECTAL CANCER

The incidence of CRC is increasing in individuals under age 50.

Recent data show a rising rate of CRC under the age of 50, despite an overall decrease in the rate of CRC diagnoses across older age groups. One in ten colorectal cancers are now diagnosed in patients younger than 50.[8] CRC is often under- and misdiagnosed in younger patients. Younger individuals are significantly more likely to be diagnosed with late stage disease compared to older individuals, due in part to delayed work-up of symptoms by the patient and/or provider.[9]

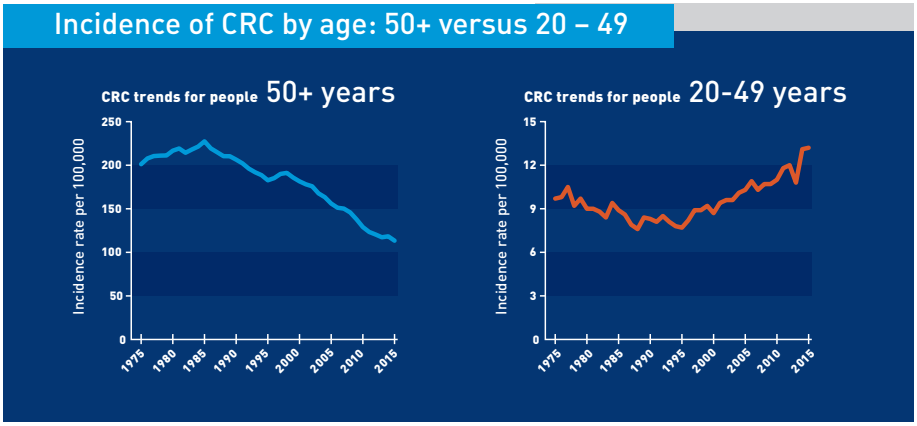

Figure 2. Incidence of CRC by age.[5]

A substantial proportion of early onset CRC may be preventable by taking a family history and screening individuals with an increased risk earlier and more frequently. Approximately 16% of cases occur in individuals with a hereditary condition, such as Lynch syndrome, and 14% have a family history of CRC.[10] Additionally, a currently undefined portion of this group has a family history of advanced adenomas that would warrant earlier screening. Early onset CRC may also develop due to personal risk factors such as chronic inflammatory bowel disease (e.g., ulcerative colitis), lifestyle factors such as limited exercise, a diet low in fruits and vegetables and high in fat, overweight and obesity, tobacco use and alcohol consumption, and other as of yet unknown causes.

In addition to routinely using family history to identify people at increased risk, primary care clinicians can help reduce CRC mortality by promoting primary prevention and early detection as well as considering CRC in the evaluation of a patient with possible alarm signs and symptoms, regardless of age.

UPDATE ON COLORECTAL CANCER SCREENING

in the general population from the American Cancer Society

The American Cancer Society (ACS) now recommends that CRC screening begin at age 45, while the US Preventative Services Task Force (USPSTF) recommended in 2016 that CRC screening should begin at age 50.[11, 12] The difference in these two recommendations is due to new data about rising incidence in younger birth cohorts that was published in 2017. See Table 1 for a comparison of the two recommendations and view an FAQ about the new guideline at NCCRT.

The ACS firmly believes that the evidence, including a concerning trend in CRC incidence in younger adults discussed in this toolkit, now points to CRC initiation starting at age 45. Having said that, ACS does anticipate that implementation will be a multi-year process, as measurement and coverage issues are worked out. ACS recognizes that many organizations will continue to follow the USPSTF recommendations for the time being. For practices that do start screening at age 45, those individuals should still be assessed for risk, as it may determine screening frequency or test selection.

Table 1. CRC screening guidelines for average risk adults: Comparison of American Cancer Society (ACS, 2018) and US Preventative Services Task Force (USPSTF, 2016) recommendations. Q = Qualified Recommendation, S = Strong Recommendation, A = A Evidence Grade, C = C Evidence Grade.

Recommendations	ACS[11]	USPSTF[12]
Age to start screening (Level of evidence)	45y Starting at 45y (Q) Screening at 50y and older (S)	50y (A)
Choice of test	High-sensitivity stool-based test or structural exam	Different methods can accurately detect early stage CRC and adenomatous polyps
Acceptable test options	FIT annually HSgFOBT annually mt-sDNA every 3y Colonoscopy every 10y CTC every 5y FS every 5y All positive non-colonoscopy tests should be followed up with colonoscopy.	HSgFOBT annually FIT annually sDNA every 1 or 3 y Colonoscopy every 10y CTC every 5y FS every 5y FS every 10y plus FIT every year
Age to stop screening (Level of evidence)	Continue to 75y as long as health is good and life expectancy 10+y (Q) 76-85y individual decision-making (Q) >85y discouraged from screening (Q)	76-85y individual decision making (C)

HOW TO USE THIS TOOLKIT

Purpose of the toolkit

The primary goal of this toolkit is to enable primary care clinicians to implement a structured family history collection system to identify individuals at increased or high risk of CRC and develop a management strategy for those individuals. A secondary goal is to facilitate timely diagnostic evaluation of patients with signs or symptoms of early onset CRC.

Learning objectives

1. Create a system to integrate family history collection and screening into practice flow
2. Identify patients at increased or high risk of CRC based on personal and/or family history
3. Apply screening guidelines to patients at increased and high risk
4. Refer high risk patients to genetic services for further evaluation, counseling, and testing
5. Include CRC in the differential diagnosis of adults under age 50 with alarm signs and symptoms

Who should use the toolkit

The toolkit is intended for primary care clinicians and administrators, including physicians, nurse practitioners, and physician assistants who specialize in internal medicine, family practice, and obstetrics/gynecology, and office managers or administrators working in these settings. Components of the toolkit may also be used by other primary care staff, such as nurses and medical assistants, who may be involved in family history collection and other associated activities.

This toolkit is designed to be used by a clinical champion or administrator to identify and implement a CRC risk assessment solution that works for the practice. The toolkit also contains guidance and education for clinicians and staff who are interested to learn more about family history collection, CRC risk assessment and risk management, and the detection of early onset CRC.

Approach towards practice change

There are different philosophies about how to introduce a new program in practice. This toolkit recommends a systemic approach with buy-in of practice or health system leadership. Other approaches could include encouraging providers and patients to engage with the program based on their interest, rather than directing a practice-wide implementation. In these cases, elements of this toolkit can still be helpful to help clinicians implement activities of interest.

Implementation and practice change are complex processes. Clinicians and staff may be able to leverage quality improvement experts from their practice or health system to assist in implementation. They may also consider additional training on evidence-based approaches that can augment the information in this toolkit. See the Appendix for select training opportunities.

Personalize the toolkit for your needs

The toolkit is designed so that you can customize your experience. Each page provides the information you need to complete a task so you can create a customized toolkit by assembling only the pages that are relevant to your practice needs.

The toolkit can be used by practices that are considering a systematic family history collection process for the first time, as well as those that may have already begun implementation who are looking for guidance on a specific issue. New and experienced users may use the toolkit in different ways. For example, practices that are new to systematic family history collection may want to read the entire toolkit prior to implementing processes, while those who have already embarked on implementation may wish to use only the tools and pages to build clinical skills around family history collection and identification of early onset colorectal cancer.

Opportunities to build on the toolkit instruction

Risk assessment beyond colorectal cancer. Recognizing that family history collection and interpretation is ideally an integrative and comprehensive process that considers risk for multiple conditions, this toolkit provides suggestions for how to implement a system for general family history collection that would allow the provider to assess a broad range of conditions. Beyond family history collection, the information about risk assessment and cancer management is specific to CRC. Practices may wish to expand their activities to include other cancers and health conditions when developing a risk assessment process.

Cancer genetic testing. Most primary care clinicians refer high risk individuals to a genetic specialist for genetic counseling and genetic testing. However, some clinicians and practices perform these processes in the primary care office, due to provider interest, patient demand, and/or limited access to genetic services. This toolkit does not provide instruction on how to integrate genetic testing into the primary care practice. Page 42 summarizes important considerations for practices considering ordering genetic testing in-house.

Navigating the toolkit in Adobe Acrobat

The toolkit contains links to external web sites and links to pages within the document. If you use internal links you may want to return to the page you were previously viewing.

You can find PDF pages that you viewed earlier by retracing your viewing path. It's helpful to understand the difference between previous and next pages and previous and next views. In the case of pages, previous and next refer to the two adjacent pages, before and after the currently active page. In the case of views, previous and next refer to your viewing history. For example, if you jump forward and backward in a document, your viewing history retraces those steps, showing you the pages you viewed in the reverse order that you viewed them.

Steps

1. Choose View > Page Navigation > Previous View.
2. To continue seeing another part of your path, do either of the following:
   - Repeat step 1.
   - Choose View > Page Navigation > Next View.

Note:
You can make the Previous View button and Next View button available in the toolbar area by right-clicking the Page Navigation toolbar and choosing them on the context menu, or choosing Show All Tools.

You can also use the keyboard shortcut. "Alt+Left Arrow" on a PC or "Command+Left Arrow" on a Mac.