RISK ASSESSMENT AND SCREENING TOOLKIT TO DETECT FAMILIAL, HEREDITARY, AND EARLY ONSET COLORECTAL CANCER

JUNE 19, 2018
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@NCCRTnews
Purpose of Today’s Webinar

• Introduce new NCCRT tool: *Risk Assessment and Screening Toolkit to Detect Familial, Hereditary, and EAO CRC.*
• Review why family history collection and timely diagnostic work ups are so critical.
• Demonstrate how the toolkit can be implemented within a system and how it will improve care for patients.
• Q&A
Presenters

Dennis Ahnen, MD
Professor Emeritus
University of Colorado School of Medicine
NCCRT Steering Committee
Co-Chair Family History and Early Age Onset CRC Task Group

Emily Edelman, MS, CGC,
Associate Director, Clinical & Continuing Education
The Jackson Laboratory

Paul Schroy, MD, MPH
Professor of Medicine
Boston University School of Medicine
Co-Chair Family History and Early Age Onset CRC Task Group

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Director of Surgical Oncology
Northwest Region at Northwell Health
NCCRT, Chair Emeritus
Co-Chair Family History and Early Age Onset CRC Task Group
Family History and Early Age Onset (EAO) CRC Task Group

Paul Schroy, MD, MPH
Family History Task Group

- Founded in 2012
- Expanded to include Early Onset CRC in 2016
- The charge is to identify key issues and areas of need around familial, inherited and early onset colorectal cancer for the purpose of identifying opportunities for the Roundtable to be a catalyst for change.
Task Group Themes

• Help clinicians develop a system-based approach to the identification and management of patients at familial risk, as well as the recommendation for early diagnostic evaluation of those presenting with signs or symptoms of CRC at any age.

• Improve EHRs to help facilitate needed screening and/or counseling recommendations for patients with a family history.

• Increase clinician-patient and intra-family communication about familial/heritable risk.

• Improve “on time” screening for the 50 to 55 population, according to recommended guidelines.

• Address the increase in CRC in young adults through strategic planning and interactions with key stakeholders and thought leaders.
Toolkit

- Jackson Lab commissioned to develop practice transformation clinician’s toolkit on family history and early onset CRC.

- Goals:
  - To bridge the existing knowledge gap and to provide a step-by-step, detailed tool for practices that are dedicated to improving their processes related to the collection of family history and acting on that information according to recommended guidelines.
  - Provide guidance on the appropriate diagnostic evaluation of patients with “alarm” signs and symptoms of CRC, regardless of age.
The NCCRT Risk Assessment and Screening Toolkit to Detect Familial, Hereditary, and Early Onset Colorectal Cancer

Emily Edelman, MS, CGC
Associate Director, Clinical & Continuing Education
The Jackson Laboratory
Toolkit Objective

Develop a system that helps primary care practices:

- Identify patients at increased/high risk based on personal and family history
- Apply screening guidelines based on risk
- Refer high risk patients to genetics
- Recognize and rapidly diagnose patients with a presenting CRC
Maya
Reason for visit
Establishing care
Medical hx
34 yo
Social hx
Married, one son
Family hx
Father with CRC @ 62
Establish a system for structured assessment

- Develop a team-based approach to family history collection and interpretation.
- Use the EHR and/or external tools to assist in family history collection and risk assessment.
- Standardize how and where family history data is recorded in the medical record to increase the usability of this information.
Stepwise instruction

**STEPS**

*For initial collection*

1. Include family history collection as a standard activity for all new patients entering the practice.

2. Determine how to best roll out your family history collection system to active patients in the practice, such as:
   - Incorporate it into preventive visits.
   - For patients that do not complete annual check-ups, run a report in the EHR to identify who has not participated and take action to include them (either through a separate appointment or adding family history collection into their next sick visit).
   - If your family history collection system does not center around an appointment with a provider, send a letter to patients and post flyers in the office advertising this new service for interested patients.

*For updating*

1. Encourage the patient to share changes to the family history over time, providing concrete examples, such as a new cancer diagnosis in a relative.

2. Update family history regularly. For adults aged 30-60 years, the family history should be updated annually in order to identify individuals that may benefit from increased cancer screening. It may be helpful to incorporate a standard question about updates to the family history during annual visits.
Implementation support and training

PARTICIPANTS
Provider, patient

WHAT YOU’LL NEED
Family history collection tool

BARRIERS
Lack of complete family history knowledge, misattributed family relationships (e.g., paternity), time

PRACTICE THIS SKILL
Web module on Collecting Family History

LEARN MORE
Selecting and Evaluating Tools for Collection and Risk Assessment

ACS Understanding Your Pathology Report: Polyps

STEPS

1. Determine which family members to interview (e.g., parents, aunts, uncles).
2. Expand to more detailed questions about cancer risk assessment if needed, and inquire about individual is more interested.
3. Ask about all known family members, including age of onset, extent of family history, and if any is multifocal or bilateral.
4. Ask of any relatives that have Crohn’s disease.
Example workflows

1. Patient arrives
2. Patient fills out FH e-questionnaire in waiting room
3. Tool calculates e-CRA
4. FHx report (PDF) imported into EHR

Provider visit:
1. Provider reviews CRA results w/ patient and determines next steps
   - High risk
   - Increased risk
   - Average risk
   - General population

Referral to cancer genetics
Provider reviews and updates patient cancer screening plan as needed
Curated tools

### Simple Family History Screening Tool for CRC

<table>
<thead>
<tr>
<th></th>
<th>YES</th>
<th>NO</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Have you had either of the following conditions diagnosed before age 50?</td>
<td></td>
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<tr>
<td>Colon or rectal cancer</td>
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<tr>
<td>Colon or rectal polyps</td>
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<tr>
<td>2. Do you have a first-degree relative (mother, father, brother, sister, or child) with any of the following conditions diagnosed before the age of 50?</td>
<td></td>
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<tr>
<td>Colon or rectal cancer</td>
<td></td>
<td></td>
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<tr>
<td>Cancer of the uterus, ovary, stomach, small intestine, urinary tract (kidney, ureter, bladder), bile ducts, pancreas, or brain</td>
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<tr>
<td>3. Do you have three or more relatives with a history of colon or rectal cancer? (This includes parents, brothers, sisters, children, grandparents, aunts, uncles, and cousins)</td>
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</tbody>
</table>

If YES to any question → Refer for additional assessment or genetic evaluation.
If NO to all → proceed with the following questions:

| 4. Do you have any first-degree relatives (mother, father, brother, sister, or child) with cancer |     |    |
Worksheets

### FAMHX TOOL FEATURES WORKSHEET

To download the spreadsheet and navigate to the tools: [https://tinyurl.com/ycqeko6h](https://tinyurl.com/ycqeko6h)

<table>
<thead>
<tr>
<th>Tool Name</th>
<th>Collection Features</th>
<th>Risk Assessment</th>
<th>Scope</th>
<th>Other</th>
</tr>
</thead>
<tbody>
<tr>
<td>Check the “must have” features for your practice:</td>
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</tbody>
</table>
| Does It Run In the Family? | YES | YES | NO | NO | n/a | n/a | n/a | n/a | n/a | YES | YES | YES | NO | YES | NO | NO | NO | NO | NO | NO | NO | NO | NO
| Family health history workbook | YES | YES | NO | YES | NO | n/a | n/a | n/a | n/a | YES | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO
| AMA adult family history form | YES | YES | NO | NO | n/a | n/a | n/a | n/a | n/a | YES | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO
| Family history questionnaire | YES | YES | NO | NO | n/a | n/a | n/a | n/a | n/a | YES | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO
| My Family Health Portrait | YES | YES | YES | PARTIAL | YES | NO | NO | NO | NO | PARTIAL | YES | YES | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO
| It Runs In My Family | YES | YES | NO | YES | NO | n/a | n/a | n/a | n/a | YES | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO
| MyLegacy | YES | YES | NO | NO | YES | YES | YES | YES | YES | NO | YES | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO
| Family healthcare | YES | YES | NO | YES | YES | YES | YES | YES | YES | NO | YES | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO
| MyTree | YES | YES | NO | YES | YES | YES | YES | YES | YES | NO | YES | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO
| Myriad family history tool | YES | YES | NO | YES | YES | YES | YES | YES | YES | NO | YES | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO
| Progeny/Kim | YES | YES | NO | YES | YES | YES | YES | YES | YES | NO | YES | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO
| CancerGene Connect/invitae | YES | YES | NO | YES | YES | YES | YES | YES | YES | NO | YES | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO
| CancerH2O | YES | YES | NO | YES | YES | YES | YES | YES | YES | NO | YES | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO
| CRIA Health | YES | YES | NO | YES | YES | YES | YES | YES | YES | NO | YES | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO
| NCI CRC Risk Assessment Tool | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO
| MMRPro | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO
| PREMIL2,6 | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO | NO

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*Note: SxX is a fictional logo.*
Example: Evaluating FH tools

● Practice’s FH goals
  o Identify patients at increased or high risk
  o Apply screening guidelines to patients based on risk level
  o Refer high risk patients to genetic services for further evaluation, counseling, and testing

● Desired FH tool features
  ✓ Structured collection of 1<sup>st</sup> and 2<sup>nd</sup> degree relatives
  ✓ Patient-entered collection
  ✓ Electronic questionnaire
  ✓ Risk assessment support
  ✓ Free
Example

- Using the FH Tool Features Worksheet, the practice can identify the tools that meet their desired needs:
  1. CancerGene Connect/Invitae
  2. CRA Health
  3. My Family Health Portrait
  4. Myriad Family History Tool
  5. Progeny/Ambry

- The practice reviews and tests the tools to select one for use.
Establish a system: additional components

- Select a set of CRC screening guidelines.
- Identify genetic and cancer specialists.
- Identify patient support materials and evidence-based interventions that can increase CRC screening adherence.
- Evaluate process and outcomes.
Assess risk

- Collect sufficient family history to enable risk assessment.
- Assess patterns and red flags.
- Assign to risk category: Average, increased (moderate or familial), high (hereditary).
Maya’s Family History

Father: 65 y/o, CRC dx 62
Paternal aunt: Died in 50s due to endometrial cancer dx 50s
Paternal uncle: 60s, gastric cancer dx 50s
Paternal cousin: 44 y/o, CRC dx 43

Colon Cancer
Gastric Cancer
Endometrial Cancer

S.E. Asian

2
50s

70
60s dx. 50s

d. 50
pneumonia
d. 70s

65
dx. 62
Hypothyroid

64

Maya
34 y

10

1

80s

80s
Identify genetic red flags and patterns

- Family history of multiple affected relatives
- Earlier age at onset of disease than expected

S.E. Asian

- d. 50
- Pneumonia

- 70
dx. 50s

- 60s
dx. 50s

- d. 70s

- 80s

- 80s

- 65
dx. 62

- 64
Hypothyroid

Maya
34 y

1

10

- Colon Cancer
- Gastric Cancer
- Endometrial Cancer

THE JACKSON LABORATORY
Colon, endometrial, and gastric cancers are associated in Lynch syndrome.
Maya is at high risk

- Multiple affected relatives
- Early age of onset
- Possible dominant pattern of associated cancers
Communicate risk & manage patient based on risk

- Develop personalized plan for cancer screening, surveillance, and prevention, and genetic referral.
- Recommend colonoscopy for increased risk individuals.
- Communicate risk levels and management recommendations.
- Tailor risk communication to patient and needs.
- Recommend patients share risk information with relatives.
- Evaluate patients with alarm signs and symptoms for CRC
Stepwise instruction

Implementation support and training

**Steps**

1. Communicate the reason for the referral. Patient may need to undergo genetic counseling if they understand.

2. Prepare your patient for what to expect during the appointment. It may seem very different compared to other medical discussions, and involvement of family members during the genetic counseling visit to help prepare the patient.

   **Tip**: For all patients and especially those that are at high risk, explain to them that genetic counseling is the process to help them understand their condition. Genetic testing is optional, and is the patient's choice. If you choose to take the test, make sure to discuss the results with your genetic counselor.

3. Provide contact information for genetic services.

   If you don't already know your local genetic professional, refer them to websites and databases which include information about geneticists:

   - National Society of Genetic Counselors (www.nsgc.org)
   - American Board of Medical Genetics (www.abmg.org)
   - International Society of Nurses in Genetics

4. Facilitate the flow of necessary information to the patient. It is effective and efficient when you can share the consent form with the patient. It may be sent to the specialist's office in advance of the appointment.

5. Schedule a follow-up to discuss the outcomes of the genetic counseling and any personalized management as indicated. Two months after the appointment, although the specific timeframe will depend on the genetic counselor.

**Participants**

Provider, patient, genetic expert

**What You'll Need**

Accessing Genetic Services Tool

**Barriers**

Lack of knowledge of where to refer, lack of patient follow-up

**Practice This Skill**

Web based module on Pre-test Decisions and Counseling

**Learn More**

Components of a GC Session Factsheet

Identifying Genetic & Cancer Specialists for Consultation

**Patient Materials**

Patient Education Materials
Tools

ACCESSING GENETIC SERVICES TOOL

Patient talking points about referral

The following points are important for you to convey to the patient in order for him or her to fully benefit from a genetic counseling appointment.

Reason for referral

Explain the reason you are referring the patient to help to set expectations and increase the likelihood of follow-through.

- Reason for referral. Some common reasons include: follow-up on family history information, discussion of risk and preventative/screening measures, assessment of appropriateness for genetic testing, or discussion of benefits and risks of genetic testing.

- Possible benefits of seeing a genetic counselor. Some benefits include: determining if you are at increased risk, determining whether genetic testing is appropriate.

- Possible harms of not pursuing the referral. Some possible harms include: not knowing about certain cancer screening or treatment options.

- Determining and communicating screening and management plans

- Summarizing and planning for follow up

  - Know that genetic testing is always optional. The appointment may or may not include genetic testing, and if it is offered, the genetic expert will discuss the benefits and risks of testing for supported decision-making.

  - Be aware testing may be recommended for affected relatives first.

  - Be aware how to prepare for the appointment. It can be helpful for patients to learn more about their family health history and to talk to affected family members about their interest and willingness to undergo genetic evaluation, in case that is recommended.

Logistics of referral

- Provide names, roles and credentials of genetic professional(s) involved
External education

CANCER PRETEST DECISIONS AND COUNSELING

Practice deciding when and if genetic testing is appropriate, given a patient's clinical and personal context.

About This Course

Communicating with patients about benefits and limitations of genetic testing is an important part of informed decision-making. In this course, you will watch a short video of a patient and provider discussion about patient motivations for genetic testing. You will practice deciding when and if genetic testing is appropriate, given the clinical and personal context and be presented with tools to help make this task easy to implement in your practice.

ACCESS CME MODULE  ACCESS CNE MODULE
Maya is a candidate for cancer genetic evaluation & testing

Referral to cancer genetics

Genetic counseling

Genetic testing for Lynch syndrome, and potentially other genes

Personalized management, prevention and surveillance
Early onset CRC is on the rise

- Recognize that the incidence of CRC is increasing in individuals under age 50.
- Be aware that a substantial proportion of early onset CRC may be prevented or detected at an earlier stage by identifying people with a family history of cancer and adenomas.
- Regardless of age, consider CRC in the evaluation of patients with alarm signs and symptoms, including:
  - blood in the stool
  - recent-onset and persistent or progressive diarrhea/constipation
  - persistent or progressive abdominal pain
  - abdominal mass
  - unexplained iron deficiency anemia
  - unexplained weight loss
A comprehensive family history and early onset CRC toolkit
Toolkit design supports applying skills and implementing processes and tools

- Step-wise instruction to help practices:
  - Establish a structured process
  - Improve clinical skills

- Tools and worksheets with worked examples

- Case studies and tips

- Appendix of guidelines, educational resources and websites

- Customizable, "build your own toolkit"
Access the toolkit

- Full comprehensive toolkit
- Quick Start version
- Web-enabled content


NCCRT Resource Center  http://nccrt.org/resource-center/
Toolkit limitations

- FH and EAO focus, not comprehensive to include all aspects of cancer genetics
- CRC focus rather than comprehensive risk assessment across conditions
- Limited evidence-based best practices
- Family history tools and EHR limitations
- Assumes organizational buy-in
- Practice variation, assumes need for some customization and modifications based on experience
Looking ahead

- Dissemination
- Implementation
  - Looking for interested practices and partners!
- Evaluation
Acknowledgements

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Potential Impact on Protecting Those At Risk

Thomas K Weber MD FACS
Cancer Family History Ascertainment in American Medical Practice is Limited – at Best.

- In an American Society of Clinical Oncology Quality Oncology Practice Initiative Survey of 10,466 (Oncology) medical records: ¹
  - Less than one-third had a complete family history. ¹
  - Less than half of medical charts documented age at cancer diagnosis.¹
  - Colorectal cancer initiated family history documentation was significantly less rigorous than that initiated by breast cancer.
  - Fewer than half of individuals with a family history of CRC or advanced adenoma (>1cm) receive personalized counseling and follow risk-based screening guidelines. ²
Identification of Those at Increased Risk

- “1 in 250 individuals have a hereditary cancer syndrome.”
- “1 in 10 individuals have increased risk for cancer based on family history.”
- “Roughly 15-20% of CRC Diagnoses occur in individuals who have at least one first degree relative with the disease.”
- A POSITIVE first degree family history for CRC increases risk 2-2.5 times.
Nearly Half of Early Age Onset CRC May Be Preventable

- “44% of Early-Onset CRC may be preventable by Family History Ascertainment linked to earlier and more frequent surveillance”

- Of 450 CRC patients diagnosed prior to age 50 in an Ohio State Study: 4
  - 16% had a named hereditary syndrome.
  - 14% had a first degree relative (FDR) with colorectal cancer.
  - 14% had a FDR with an advanced adenoma.”
Identifying Individuals at **Significantly Increased Risk of Breast Cancer**

- One or more First or Second degree relatives with:
  - Early age onset Breast Cancer (< 45 years).
  - Male breast cancer
  - Ovarian cancer
  - Breast and ovarian cancer in the same relative

- Genetic counseling and testing recommended
Additional life threatening but manageable / treatable conditions associated with positive family history:

- Coronary Artery Disease
- High Blood Pressure
- Blood Clotting Disorders
- Diabetes
- Asthma
- Prostate Cancer
Risk Assessment and Screening Toolkit

*Functional Electronic Medical Record Clinical Decision Support Essential to Success!*

Thomas K Weber MD FACS
Risk Assessment and Screening Toolkit

“Don’t Live in Mystery; Know Your Family History!”™

TM : tkw
REFERENCES

1. Wood et al JCO 2014 32(8) 824

2. NCCRT Risk Assessment Tool Kit 2018

3. Lowery et al Cancer 2016 122(17) 2633-2645

4. Pearlman & Hampel et al JAMA Onc 2017 3(4) 464-71

5. CDC – on line references
Please submit your questions in the chat box.
Thank You!

To follow NCCRT on social media:
- Twitter: @NCCRTnews
- Facebook: www.facebook.com/coloncancerroundtable

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