GECKO
Genetics Education Canada: Knowledge Organization
Centre d'éducation en génétique canadien: Connaissances organisées

POINT OF CARE TOOL

PART 1: COLORECTAL CANCER RISK ASSESSMENT TOOL TO IDENTIFY INDIVIDUALS MOST LIKELY TO BENEFIT FROM REFERRAL TO GENETICS

Updated Jan 2023

1 Do you have a first-degree relative (mother, father, brother, sister, or child) with any of the following conditions diagnosed before age 50:

- ☐ Colon or rectal cancer?
- ☐ Cancer of the uterus, ovary, stomach, small intestine, urinary tract (kidney, ureter, bladder), bile ducts, pancreas, or brain?

2 Have you had any of the following conditions diagnosed before age 50:

- ☐ Colon or rectal cancer?
- ☐ Colon or rectal polyps?

3 Do you have three (3) or more relatives with a history of colon or rectal cancer?
include parents, brothers, sister, children, grandparents, aunts, uncles, and cousins

☐

If an individual answers "yes" to all of these questions a referral to genetics should be offered. If a patient answers "yes" to any of these questions, consider further assessment using the criteria in Part II.

The cumulative sensitivity of these three questions to identify patients with characteristics suggestive of hereditary colorectal cancer and who should undergo a more extensive risk assessment is 77%. When all 3 questions were answered "yes", the tool correctly identified 95% of individuals with pathogenic gene variants causing Lynch syndrome.

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PART II: RED FLAGS TO IDENTIFY INDIVIDUALS AT HIGH RISK FOR LYNCH SYNDROME MOST LIKELY TO BENEFIT FROM A GENETIC ASSESSMENT

Updated Jan 2023

LYNCH SYNROME RELATED CANCERS

Colorectal

Endometrial

Ovarian

Gastric

Small bowel

Urinary tract

Hepato-biliary

Brain, mainly glioblastoma

Skin (sebaceous adenoma or carcinoma)

Pancreatic

PERSONAL HISTORY

Strongly consider referral for genetic assessment where:

Lynch syndrome-related cancer (*see above*) AND

- Tumour pathology supporting a diagnosis of Lynch syndrome e.g. high microsatellite instability (MSI), immunohistochemistry (IHC) showing loss of expression of a Lynch syndrome gene (*MLH1, MSH2, MSH6,* and/or *PMS2*), genomic testing showing a pathogenic variant in a Lynch syndrome gene

Colorectal or endometrial cancer diagnosed at age ≤50 years **AND**

- A second Lynch syndrome-related cancer **OR**
- One or more first-degree relative(s) with any Lynch syndrome-related cancer diagnosed at age ≤50 years **OR**
- Two or more first-degree relatives with any Lynch syndrome-related cancer diagnosed at any age

FAMILY HISTORY

Strongly consider referral for genetic assessment where:

There is a known pathogenic or likely pathogenic variant in a Lynch syndrome gene (*MLH1, MSH2, MSH6, PMS2, EPCAM*)
Documentation such as a test report or family letter is ideal and may be required for testing

Three (3) or more relatives were diagnosed with a Lynch syndrome-related cancer (*see above*) **AND** the following criteria are met:

- Two (2) of the affected individuals are first-degree relatives
  i.e. parent and offspring, siblings
- Two (2) or more successive generations are affected
  suggesting autosomal dominant inheritance
- One (1) or more cancer diagnoses were diagnosed at age ≤50 years
  Verification of diagnosis with tumour pathology where possible
  From revised Amsterdam criteria

A risk prediction model estimates ≥5% probability of carrying a pathogenic variant in a Lynch syndrome gene
e.g. PREMM5, MMRPredict, MMRPro

For more information on Lynch Syndrome such as screening recommendations see the complete GECKO Messenger at www.geneticseducation.ca