CANCER COLORECTAL CANADA | COLORECTAL CANCER CANADA

THE DON MCQUAIG FOUNDATION Get Personal UNLOCK YOUR TREATMENT OPTIONS

HEREDITARY COLORECTAL CANCER SYNDROMES, BIOMARKER TESTING, SCREENING AND PREVENTION

Did you know having a family health history of colorectal cancer increases your risk to get colorectal cancer yourself?

Knowing your family health history of colorectal cancer and sharing this information with your doctor can help you take steps to lower your risk and create a personalized screening program.

If you have a family health history of colorectal cancer, your doctor may recommend the following screening methods for prevention and/or early detection:

- start screening at a younger age
- create a personalized screening plan
- get colonoscopies and other diagnostic testing
- In some cases, receive genetic counseling

What are the different colorectal cancer hereditary syndromes?

Hereditary cancer syndromes are caused by mutations (changes) in certain genes passed from parents to children.

LYNCH SYNDROME

- Also known as hereditary nonpolyposis colorectal cancer (HNPCC), an inherited condition that increases the risk of colon cancer, endometrial cancer and several other cancers.
- The most common hereditary colorectal cancer syndrome (accounts for 2% to 4% of all colorectal cancers)
- Caused by an inherited mutation in either the MLH1, MSH2 or MSH6 gene, or or high-level microsatellite instability (MSI-H)

FAMILIAL ADENOMATOUS POLYPOSIS (FAP)

- Characterized by numerous adenomas (benign tumours of the glands) in the rectum and colon leading to colorectal cancer
- Causes less than 1% of colorectal cancer cases
- Attenuated familial adenomatous polyposis (AFAP) is a milder variant of FAP and presents with less colorectal polyps (<100) and later presentation of adenomas and colorectal cancer.
- Both FAP and AFAP are caused by mutations in a tumour suppressor gene called Adenomatous polyposis coli (APC) gene.

Certain other inherited syndromes, such as MUTYH-associated polyposis and Peutz-Jeghers syndrome, can also greatly increase a person's risk of colorectal cancer.

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15%

of colorectal cancer tumors are microsatellite instability-high (MSI-H) or mismatch repair deficient (dMMR).

Why should I receive biomarker testing?

If you are diagnosed with cancer, biomarker testing may enable the selection of the most appropriate treatment for you.
For example:

- Detection of MSI-H dMMR is predictive of Lynch syndrome. MSI-H patients are considered candidates for targeted therapy or immunotherapy (Pembrolizumab (KEYTRUDA)).
- Germ-line APC mutations are considered an early detection marker because nearly 100% of individuals with the mutation will develop colon cancer in the future.

When should I get my biomarkers tested?

The best time to do testing is soon after a colorectal cancer diagnosis and before a treatment plan is chosen. Specifically, if you are diagnosed with stage IV metastatic colorectal cancer, you should be tested for at least three predictive biomarkers: RAS, BRAF, and MSI/dMMR.

According to the Bethesda guidelines, you should be tested for MSI/dMMR in the following situations:

Colorectal cancer diagnosed at age younger than 50

Presence of synchronous, metachronous colorectal, or other HNPCC-associated tumors, regardless of age.

Colorectal cancer diagnosed and MSI-H histology diagnosed by age 60

Colorectal cancer diagnosed in two or more first- or second-degree relatives with HNPCC-related tumors, regardless of age.

Colorectal cancer diagnosed in one or more first-degree relatives with an HNPCC-related tumor, with one of the cancers being diagnosed under age 50 years.

More information on biomarkers and biomarker testing can be found in the pamphlet "Colorectal Cancer and Biomarker Testing - What Patients Need to Know" here: www.colorectalcancercanada.com >>What We Do >> Our Programs >> Get Personal