

Referral Guidance for Hereditary Cancer Genetic Assessment



The full eligibility criteria and gene panels for hereditary cancer testing (HCT) can be found at cancercareontario.ca/en/guidelines-advice/types-of-cancer/70161. Genetic testing is most informative when first performed for an individual with a personal history of cancer - refer them first if possible. A genetic assessment can determine test eligibility and guide appropriate test selection and interpretation. A genetics clinic will ultimately determine if an appointment and testing is indicated.

Who should be referred for genetic assessment?

- ☐ **KNOWN FAMILIAL VARIANT:** Confirmed hereditary pathogenic/likely pathogenic variant in a blood relative¹
- ☐ **YOUNG** age at diagnosis, such as:
 - Age 18 or younger for any cancer
 - Age 45 or younger for renal cancer
 - Age 50 or younger for breast, prostate, colorectal, endometrial, gastric, gastroesophageal junction cancers
- ☐ **MULTIPLE** primary cancers in an individual
- ☐ **MULTIPLE** of the same or related* cancers on the same side of a family¹, such as:
 - breast and ovarian cancer
 - colon and endometrial cancer
 - melanoma and pancreatic cancer

** For additional details on cancer associations within specific hereditary cancer syndromes, see the full HCT guidance linked above.*
- ☐ **SPECIFIC OR RARE DIAGNOSIS**, such as any of the following cancers:
 - Pheochromocytoma or paraganglioma
 - Ovarian cancer
 - Male breast cancer
 - Triple negative breast cancer
 - High-risk or metastatic prostate cancer²
 - Pancreatic cancer
 - Biliary tract cancer
 - Abnormal mismatch repair (MMR) immunohistochemistry (IHC) (pathology results suggestive of Lynch syndrome)
 - Multiple adenomatous gastrointestinal polyps³ (i.e., ≥10 polyps at ≤60 years; ≥20 polyps at any age)
- ☐ **ANCESTRY** associated with higher cancer risk:
 - Ashkenazi Jewish ancestry with breast, prostate, colorectal cancer or GI polyps
 - Māori ancestry with diffuse gastric cancer

What are the next steps?

If a referral is appropriate, use the [Ontario Genetics Clinic Directory](#) to find your local clinic that is suited to your patient (e.g., adult vs. pediatric). Submit a detailed referral along with any prior related reports.

Ask your patient to gather relevant information, including:

- ☐ Age at diagnosis AND type of cancer in relative(s) with as many details as possible (e.g., clarifying if a gynecologic cancer in a family member was cervical, endometrial, or ovarian)
- ☐ Relatives' pathology and/or genetic test reports or family letter (if available)

Still have questions? Consider consulting with a geneticist via the [Ontario eConsult](#) program. For general assistance navigating genetic services contact a genetic counsellor through [gcConnect](#) (1-844-564-4363 or gcConnect@ontariohealth.ca). Do not share PHI with gcConnect. A comprehensive list of provincial cancer resources and programs is on the Ontario Health website: ontariohealth.ca/clinical/cancer

¹ Typically includes first- and second-degree relatives; exceptions may include limited or uninformative family history (e.g., adoption, no contact with family, small family size, all brothers or all sisters depending on cancer type, etc.)

² One or more features: T3 (or higher) staging, Grade Group 4 or 5, lymph node involvement, PSA ≥20

³ Consider referring patients suggestive of other colorectal polyposis conditions based on the full HCT eligibility criteria