

Cancer Genetics Referrals: When to Refer to Genetics

- Genetic risk assessment and testing can be helpful in aiding in the care of individuals with a personal or family history of cancer.
- A genetic predisposition to developing cancer can be inherited through a **mother or father** for both **males and females**.
- Those with an unknown or limited family history can still be referred to genetics for evaluation.
- Genetic test results can:
 - Impact treatment recommendations
 - Aid in surgical management decision making
 - Inform unaffected individuals of future risks
 - Guide appropriate screenings

General Referral Guidelines for Cancer Indications

Individuals with any of the following in their **personal or family history**:

- A **known genetic pathogenic variant/mutation identified** in a cancer susceptibility gene
- **Any** of the following **diagnosed at any age**:
 - Ovarian cancer (including fallopian tube and primary peritoneal cancers)
 - Male breast cancer
 - Pancreatic cancer
 - Metastatic prostate cancer
 - Medullary thyroid cancer
 - Pheochromocytoma/Paraganglioma
 - Adrenocortical carcinoma
- **Any** of the following **diagnosed at a young age**:
 - Breast cancer
 - Colorectal cancer
 - Endometrial cancer
 - Gastric cancer
 - Other cancer(s) diagnosed at unusually young ages
- **Two or more** cancers diagnosed **in the same person**
- A personal and/or family history of **≥ 3 cancers on the same side of the family**
- Cancers with **specific tumor characteristics** reported from pathology:
 - Triple negative breast cancer
 - Colon, uterine or other cancers showing evidence of mismatch repair (MMR) deficiency, either by microsatellite instability (MSI) or loss of MMR protein expression at any age
- Individuals of **Ashkenazi Jewish descent** with a personal or family history of breast, ovarian, pancreatic, or high-grade prostate cancer
- **Non-cancerous personal or family history** that may indicate referral include:
 - ≥ 10 cumulative adenomatous polyps
 - ≥ 2 hamartomatous polyps
 - ≥ 5 serrated polyps proximal to sigmoid colon
 - Parathyroid, pituitary, or carcinoid tumors
 - Dysmorphic features in addition to cancer history
 - Mucocutaneous hyperpigmentation of the mouth, lips, nose, eyes, genitalia, or fingers

Full referral guidelines are referenced on the back of this handout. If you are concerned about a patient's personal or family history of cancer, a genetics professional can help determine if further risk assessment and/or genetic testing is indicated.

Cancers Associated with Common Hereditary Cancer Syndromes

Hereditary Breast and Ovarian Cancer (HBOC) Syndrome

- Breast cancer
- Ovarian cancer
- Prostate cancer
- Pancreatic cancer
- Sarcoma
- Adrenocortical carcinoma
- Thyroid cancer
- Brain tumor
- Leukemia
- Colorectal cancer
- Endometrial cancer
- Kidney cancer
- Diffuse gastric cancer
- Mucocutaneous skin lesions

Lynch Syndrome

- Colorectal cancer
- Endometrial cancer
- Gastric cancer
- Ovarian cancer
- Pancreatic cancer
- Ureter/renal pelvis cancers
- Biliary tract cancer
- Brain cancer (usually Glioblastoma)
- Small intestinal cancers
- Sebaceous adenomas
- Sebaceous carcinomas
- Keratoacanthomas

Take Action

If your patient meets any of these criteria:

- Talk to your patient about recommendations for a genetics referral
- For assistance locating the nearest genetics service provider, in the New York-Mid-Atlantic Consortium (NYMAC) Region, please contact the **Genetic Services Referral Call Center** at **1-833-545-3218** or visit our [website](#).

Guidelines and Recommendations (Links)

- **HEREDITARY BREAST/OVARIAN CANCER**
[National Comprehensive Cancer Network \(NCCN\) Guidelines for Genetic/Familial High-Risk Assessment: Breast and Ovarian](#)
[US Preventative Services Task Force \(USPSTF\) Recommendations for BRCA-Related Cancer](#)
- **HEREDITARY COLORECTAL CANCER/POLYP SYNDROMES**
[National Comprehensive Cancer Network \(NCCN\) Guidelines for Genetic/Familial High-Risk Assessment: Colorectal Evaluations of Genomic Applications in Practice and Prevention \(EGAPP\) through the CDC](#)
- **MULTIPLE CANCERS**
[National Society of Genetic Counselors/American College of Medical Genetics and Genomics \(NSGC/ACMG\) Referral Indications for Cancer Predisposition Risk Assessment](#)

Resources (Links)

- [National Society of Genetic Counselors \(NSGC\) Find a Genetic Counselor](#)
A tool developed by NSGC for patients and providers to locate genetic counseling services in North America (U.S. and Canada).
- [American Cancer Society \(ACS\)](#)
Provides resources regarding cancer diagnosis, treatment, research, support services, and support resources.
- [American Society of Clinical Oncology \(ASCO\)](#)
Professional organization for oncology professionals with resources for providers and patients.
- [American College of Obstetricians and Gynecologists \(ACOG\)](#)
Non-profit professional organization with guidelines and educational resources for improving women's health.
- [GeneReviews through the NIH](#)
Resource for providers about the diagnosis and management of patients with genetic conditions.



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