UT Southwestern
Simmons Cancer Center

Genetic Testing Information for the Primary Care Provider

Impact of Genetic Testing on Patient Care

10% of cancer occurs due to an inherited mutation. Hereditary cancer predisposition syndromes increase the risk for various cancers and can be passed down from generation to generation. The primary goal of cancer genetic testing is to help identify individuals and families that may be at increased risk for hereditary cancer.

Individuals who have a hereditary cancer predisposition syndrome may need more frequent cancer surveillance or cancer management options (such as a prophylactic surgery) to help manage their risks.

Cancer predisposition syndromes are severely underdiagnosed. Taking a family history is the first step to identify individuals that may need further evaluation. This should include cancer histories from first-degree (children, siblings, parents), second – degree (grand-parents/-children, aunts/uncles, nieces/nephews) and third-degree (cousins, great-grandparents/-grandchildren, great-nieces/-nephews) relatives.

Hallmark Features of Hereditary Cancer

- Multiple family members with the same or related types of cancer (e.g. breast and ovarian; colon and uterine)
- Cancer in multiple generations
- Cancer occurring at a young age (<45-50 years)
- Bilateral or multiple primary tumors
- Rare cancers (male breast cancer)

When to Refer to Cancer Genetics

Common referral indications are described in the table below. Please note that this list is not all-inclusive and may change with time. The American College of Medical Genetics and Genomics and the National Society of Genetic Counselors have complete practice guidelines for referring to cancer genetics, which can be found at www.ACMG.net and www.NCCN.org. Your local genetics office can also be a resource for answering referral questions.

Common Referral Indications

Breast Cancer	Colon Cancer	Ovarian Cancer	Pancreatic Cancer	Prostate Cancer	Uterine Cancer
One breast cancer in patient/family if: • Dx ≤50 • Dx ≤60 if triple negative • Two relatives with prostate, ovarian, or pancreatic cancer • Ashkenazi Jewish • Male **Two breast cancers in patient/family if:** • At least one dx <50 **Three breast cancers in patient/family at any age**	**One colon cancer in patient/family if:** • Dx < 50 • Same person has second Lynch syndrome cancer* • Relative has Lynch syndrome cancer* dx <50 • Two relatives have Lynch syndrome cancer*	**One ovarian cancer in patient/family at any age**	**One pancreatic cancer in patient/family at any age**	**One prostate cancer in patient/family if:** • Metastatic • Ashkenazi Jewish • Relative with ovarian cancer • Relative with male breast cancer • Relative with pancreatic cancer • Relative with breast cancer dx ≤50 • Two relatives with breast cancer **Three prostate cancers in family at any age**	**One uterine cancer in patient/family if:** • Dx < 50 • Same person has second Lynch syndrome cancer • Relative has Lynch syndrome cancer dx <50 • Two relatives have Lynch syndrome cancer

Dx = diagnosed

*colon, uterine, gastric, pancreatic, ovarian, ureter/renal pelvis, biliary tract, small intestine, brain, sebaceous adenoma/carcinoma, keratoacanthoma

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Current Genetic Testing Options for Hereditary Cancer

Hereditary cancer genetic testing now includes many options consisting of panels that assess multiple genes at once. There are many options for panels depending on patient preference and personal/family history. Various commercial testing laboratories offer smaller, tailored panels for common cancers such as breast cancer and colon cancer, while other options include larger panels that test for a wide range of hereditary cancer indications.

Insurance and Financial Options

Most insurance companies cover the cost of genetic testing for patients who meet certain criteria, and financial assistance may be available for patients without insurance. Competitive self-pay options are also available.

Criteria for genetic testing varies greatly depending on the type of cancer, but generally includes patients with a personal/family history of young diagnoses (often <45-50 years old), multiple primary cancers in one person, and multiple family members with the same or related types of cancers (e.g. breast and ovarian).

How can I learn more?

For information about genetic testing and genetic counseling, visit the UT Southwestern Cancer Genetics for Health Professionals site.

For additional resources on coordinating genetics counseling and testing for patients, or providing genetics services, visit the Genetic Screening and Navigation Toolkit.

To find a genetic counselor near you, visit the National Society of Genetic Counselors Find a Genetic Counselor Tool.

Resources

Hampel H., Bennett R., Buchanan A., Pearlman R., Wiesner G. A practice guideline from the American College of Medical Genetics and Genomics and the National Society of Genetic Counselors: referral indications for cancer predisposition assessment. *Genet Med*. 2014. 17(1):70-87.

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