UT Southwestern
Simmons Cancer Center

Genetic Testing Information for Gastroenterologists

Impact of Genetic Testing on Patient Care

Genetic testing for hereditary cancers can be a useful tool for affected patients as well as their family members. It can also be helpful for unaffected patients with a significant family history of cancer.

For patients considering surgical options, genetic testing can help patients make informed decisions regarding their healthcare. For example, patients who test positive for a pathogenic variant in a cancer-related gene may elect to undergo a more extensive surgery.

Additionally, patients who test positive may be at an increased risk for other related cancers (e.g. uterine cancer or prostate cancer). Genetic testing may help clarify these risks and allow patients to consider increased cancer screenings or even take preventative measures, such as prophylactic surgery.

Family members of patients who test positive are recommended to pursue genetic testing as well as consider increased cancer screening as appropriate.

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 y)
- Bilateral or multiple primary tumors
- Rare cancers (e.g. male breast cancer)

When to Refer to Genetic Counseling

Common referral indications are outlined 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 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 for genetic counseling you may come across in your clinic include:

- A personal history of colorectal cancer diagnosed < 50
- A personal history of colorectal cancer with a tumor showing evidence of mismatch repair (MMR) deficiency or microsatellite instability (e.g. MSI-high)
- A personal history of colorectal cancer and another Lynch Syndrome-associated tumor (stomach, small intestine, endometrium, ovary, ureter/renal, brain, biliary tract, or pancreas)
- A personal history of pancreatic cancer
- A first degree relative diagnosed with pancreatic cancer
- A personal history of diffuse gastric cancer diagnosed < 40
- A family history of two or more gastric cancers with at least one being diffuse type
- A personal or family history of diffuse gastric cancer and lobular breast cancer, with one being diagnosed < 50
- A personal history of > 10 adenomatous polyps, multiple hamartomatous polyps, ≥ 5 juvenile polyps in the colon, ≥ 5 serrated polyps proximal to the sigmoid colon
- A family history of three or more GI cancers or Lynch syndrome-associated cancers not in the GI system (endometrium, ovary, ureter/renal, brain)

Current Landscape of Testing for Hereditary Cancer

Commercial genetic testing laboratories now offer hereditary cancer genetic testing panels, which assess multiple genes at once and test selection is often guided by patient preference and personal/family history. Update genetic testing should be considered for patients who underwent testing several years ago (typically prior to 2014).

Insurance and Financial Options

Most insurance companies cover the cost of genetic testing, and financial assistance may be available for patients without insurance. Self-pay options are also available.

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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.