

Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process integrates interpretation of family and medical histories to assess the chance of disease occurrence or recurrence; education about inheritance, testing, management, prevention, resources and research; and counseling to promote informed choices and adaptation to the risk or condition.

The Saint Joseph Mercy Cancer Genetics Program includes board certified physicians in internal medicine and medical oncology, two genetic counselors, one who is board certified and one who is board eligible, and support staff. The program provides risk assessment and genetic counseling to patients with a personal and/or family history of cancer in accordance with the Commission on Cancer Standard 2.3. Referral by a physician or advanced practitioner is required. Upon referral, patients are given a 10-page questionnaire to collect information about their screening practices, medical history, family history, and ancestry/ethnicity. The genetic counselor uses this information to create a 3-generation pedigree and complete a risk assessment using established models such as BRCApro, MMRpro, PREMM 1,2,6, etc. The genetic counselor also determines whether the patient meets the NCCN guidelines for genetic counseling/testing and verifies insurance coverage.

At the pre-test appointment, the patient meets with a genetic counselor for education about his/her personal risk assessment and suspected hereditary cancer syndrome(s). This includes a discussion about genes and inheritance patterns, cancer risks associated with gene mutations, medical management options for high risk individuals, benefits and risks of genetic testing, and possible test results. A psychosocial assessment is performed, and further counseling is provided as needed. If the patient is interested in genetic testing, the genetic counselor obtains informed consent, completes appropriate paperwork and coordinates a blood draw. If the patient's insurance requires prior authorization, the genetic counselor is responsible for obtaining authorization and sharing it with the testing laboratory. A consult note is sent to the referring physician and the patient following the appointment.

The genetic counselor will disclose and explain genetic test results to the patient via telephone and determine if follow-up appointments are necessary. An in-person follow-up appointment with a genetic counselor is not required if the patient tests negative (i.e., no mutation detected), although post-test counseling via phone is provided. If the patient is part of the Saint Joseph Mercy High Risk Breast Program, an appointment with a designated physician is coordinated to review management options based on the genetic test result, family history and personal factors. When testing identifies a pathogenic gene mutation or variant of unknown significance, post-test appointments are coordinated in one of two ways:

- For patients who do not have an established medical oncologist:
  - The patient meets with their genetic counselor and a designated medical oncologist to discuss the significance of the results, describe available medical management options and make necessary referrals, and review the inheritance pattern and importance of informing relatives of the test results. In some cases, the medical oncologist recommends further follow-up in her office.
- For patients with an established medical oncologist:
  - The genetic counselor explains the significance of the result, describes available medical management options, and reviews the inheritance pattern and importance of informing relatives of the test results. The genetic counselor will recommend that the patient follow-up with their established medical oncologist to review implications and make necessary referrals.

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When testing identifies a gene mutation, the genetic counselor also provides the patient with an informational letter to share with relatives and a packet of resources for further information and support. When testing identifies a variant of unknown significance, the genetic counselor will discuss available research opportunities and help coordinate family studies as needed. Upon disclosure and explanation of the genetic test result, a consult note and copy of the test report are sent to the patient and referring physician following the appointment.

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