

# **Clinical Genetics Service**

Identifying inherited cancer risk and explaining options for managing and lowering that risk



Roswell Park's Clinical Genetics Service assists patients whose personal or family medical histories suggest an increased risk of cancer due to an inherited genetic mutation.

www.roswellpark.org/partners-in-practice

### **Red Flags for Referral**

Only about 5-10% of cancers are linked to inherited genetic mutations, so genetic testing is not recommended for everyone. Patients may be candidates for referral if:

- They have several first-degree relatives (parents, siblings, children) who have had cancer.
- Their family medical history reveals multiple relatives who have had: — the same types of cancer
  - multiple primary cancers, or
  - cancers that tend to cluster together (e.g., breast/ovarian).
- Their family tree includes multiple relatives who were diagnosed with cancer before age 50.
- They are of Ashkenazi Jewish ancestry or come from other ethnic groups that may have a higher risk for certain hereditary cancer syndromes.

## **Partners In Practice**

medical information for physicians by physicians

## Multi-Gene Panels:

The Importance of the Genetic Counselor

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#### **Professional organizations**

such as the National Comprehensive Cancer Network and the Society for Gynecologic Oncology have begun to recommend that, due to their intrinsic complexity, multi-gene panels be ordered only in consultation with a genetics professional. Genetic counselors are readily available to help both patients and providers. Within the past year, the development of hereditary cancer multi-gene panels has led to changes in genetic testing. The panels use next-generation sequencing to analyze several different genes simultaneously, saving time and money.

#### However, several factors can complicate the interpretation of the test results, including:

- The presence of moderate-risk genes
- A high rate of variants of unknown significance
- Limited medical-management guidelines

Consequently, testing through multi-gene panels should be considered only in the presence of a trained genetics professional after pre-test counseling and completion of an individualized assessment of the patient's personal and family medical history.

#### Not all laboratories' genetic testing panels are created equal.

- Panels vary in terms of the genes included and how thoroughly each gene is evaluated. A panel with more genes may not be better than a smaller panel.
- Some panels include genes about which very little is known; a mutation in these genes may not impact medical management.
- Panels vary in their ability to detect mutations (sensitivity) due to differences in the technology used to find and confirm mutations.

Genetic counselors stay abreast of ongoing updates to laboratory panels. The genetic counselor's knowledge and expertise allow for identification of the best panel based on the patient's personal and family history.

#### Panel tests often identify variants of unknown clinical significance.

- A variant of unknown clinical significance (VUS) is a gene change for which there is limited information to know whether it is a benign polymorphism (common gene change) or a true mutation.
- A VUS can be difficult for a provider to manage and for a patient to understand.
- Laboratories vary in their rates and methods of reporting and classifying variants. They also differ regarding whether they update providers when a VUS is reclassified.

Genetic counselors use their expertise evaluating the pertinent history and available scientific data regarding the nature of variants to ensure appropriate utilization of test results. They can assist patients and family members with understanding the complexities of a VUS and update them regarding new interpretations over time.

#### A panel test may or may not always be the most cost-effective test.

- It is important to conduct a complete risk assessment (including family history analysis) before choosing a multi-gene panel. Patients may require only a single-gene test for significantly less cost.
- When multiple genetic etiologies are in the differential diagnosis, a panel may save time and money.
- The cost of panels varies widely from lab to lab, as do billing policies, turnaround time, and reporting practices.

The inclusion of genetic counselors within a clinical practice allows for the most appropriate utilization of valuable health-care dollars.

## Roswell Park's Clinical Genetics providers can:



• Provide an individualized cancer genetic risk assessment

to gauge the likelihood of hereditary cancer risk in a family; identify hereditary cancer syndromes in the differential diagnosis, as well as the most appropriate genetic testing approach to evaluate for those syndromes; and determine which individuals in a family are the most appropriate candidates for genetic testing.

- Educate the patient and family members about the biology and genetics of cancer, and discuss the social, ethical, and legal issues related to genetic testing.
- Arrange for genetic testing via a blood test performed off-site.
- Explain the results of genetic tests.
- Explain options for managing cancer risk, if indicated. These may include more frequent cancer screening or initiating screening at an earlier age; risk-reducing surgery (e.g., thyroidectomy or mastectomy/oophorectomy); or chemoprevention.

## Periodic follow-up is essential.

The past 10 years have led to significant improvements in assessing hereditary cancer risk, due largely to advancements in the technology of genetic testing, knowledge of hereditary cancer risks, and identification of new hereditary cancer-susceptibility genes. With this expansion of information and resources, many patients who in the past were not eligible for genetic testing may now be candidates. At the same time, patients who were tested previously and found to have no mutation(s) may now be eligible for additional testing of the same gene(s) or may be candidates for evaluation for other hereditary cancer syndromes.

Patients suspected of having a hereditary risk for cancer should be referred, or re-referred, to a genetics provider for formal evaluation and possible facilitation of genetic testing. If you are not sure whether referral is appropriate for a patient, please call us at 716-845-8400.

## Meet the Clinical Genetics Team



- Bonnie Braddock, MPH, CGC
- Genetic Counselor
  2 Mollie L. Hutton, MS, CGC
  - Genetic Counselor





Icoleta Voian, MD, MPH Director, Clinical Genetics Service

## **Patient Referrals**

Patients may call RPCI's Cancer Information Program directly, at **1-877-ASK-RPCI** (1-877-275-7724). An information specialist will complete a brief genetics questionnaire with the patient over the phone, and the patient will be contacted later about an appointment.





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