Genetic Risk Evaluation and Testing

Many people have a family member who has had cancer. While the majority of cancer happens by chance, about 5-10 percent of people with cancer have an inherited cancer. Inherited refers to a change in a gene that runs in families and may be passed down from parents to children, causing a higher risk for cancer. It is important to know which individuals have an inherited cancer risk so that they can take steps for early detection or preventative measures to reduce the risk of cancer and save lives.

Why are some cancers inherited?

All cancers are caused by a change or mutation in a gene. Genes are like blueprints for the body or instructions our body uses to grow and function. When a gene mutation or change occurs, genes do not function correctly, and this may cause a higher risk for cancer. You have two copies of every gene, one from each parent. Mutated genes that are passed down from parent to child increase the risk for certain cancers. You can inherit a gene change or mutation from either parent.

Factors Associated with Hereditary Cancer

- Multiple cancers in one person
- Cancer diagnosed at a young age
- Multiple family members with the same type of cancer
- Cancer in multiple generations

- Rare cancers or in the gender not usually affected (like male breast cancer)
- Cancer in paired organs (both breasts, ovaries, kidneys)
- More than one childhood cancer in siblings

What is genetic testing or a genetic risk evaluation?

A genetic risk evaluation is done by a trained medical professional to determine your risk for developing cancer, appropriate medical management, and make genetic testing recommendations. A genetic evaluation includes:

- **Medical History:** A detailed review of your personal medical and family history. This information helps determine your risk of developing cancer, appropriate medical management, and if genetic testing is recommended to help clarify your cancer risk.
- **Testing:** Genetic testing is an analysis of a person's genes (usually through a blood sample) to determine if you have a change in a gene, called a mutation, that increases the risk for cancer. Insurance companies usually cover the cost of testing if you have a personal or family history that is concerning for hereditary cancer. Cost and insurance coverage for testing are discussed during your evaluation.
- Counseling: Following the testing, the person receives comprehensive counseling based on the results of the test and family history. For those who are found to have a gene mutation or a higher risk of cancer, options for next steps are discussed.
- Next Steps: Options can include surveillance or prevention strategies. A patient's choice is strictly a personal decision. Genetic counseling and testing provide the tools needed to make informed decisions.
- Family Risk: If a family member has tested positive for a mutation, testing of other family members is encouraged. The information from genetic counseling and testing enables family members to make decisions that could save their lives. It also can tell family members that they do not have the mutated gene.

Benefits of Genetic Testing	Risks of Genetic Counseling and Testing
Relief from uncertainty	• Difficulty coping with cancer risk with limited information
Understand your cancer risk	• Impact on family and personal relationships
Make informed medical and lifestyle decisions	Concern about privacy
Provide helpful information for relatives	• Cost

Confidentiality of Results

The results of genetic counseling and testing are strictly confidential. It is against the law for health insurance companies to discriminate against someone based on genetic information (GINA Law). The law does not apply to life, disability, or long-term care insurance; very small companies; or many federal health plans.

Hereditary Breast and Ovarian Cancer Syndrome **Lynch Syndrome Risk Factors Risk Factors** • Breast cancer at age 45 or younger Lynch syndrome related cancers include: colon, • Ovarian, male breast, pancreatic, intraductal prostate cancer, endometrial (uterine), ovarian, stomach, ureter/renal pelvis, metastatic prostate, metastatic (HER2 negative) breast cancer biliary tract, small bowel, pancreas, brain, or Sebaceous adenomas • 2 breast cancers in a person, with 1 before age 65 years • Colon or endometrial cancer before age 50 • Triple negative breast cancer at age 60 or younger • A person with 2 or more of the above Lynch cancers • 2 or more relatives with breast cancer with 1 before age 50 • 2 or more relatives with a Lynch cancer, with one • 3 or more relatives with breast, ovarian, pancreatic and/or diagnosed before the age of 50 aggressive prostate cancer at any age • 3 or more relatives with a Lynch cancer • Ashkenazi Jewish ancestry • Known family mutation in a Lynch Syndrome gene • A relative with a mutation in BRCA1 or BRCA2

Sources: American Cancer Society, American Society of Clinical Oncology, National Cancer Institute, National Comprehensive Cancer Network, National Institutes of Health

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