A biomarker test analyzes tissue or blood samples for the presence of biomarkers that can provide insights into diseases like cancer. Medical treatment can be guided by biomarker testing, often referred to as precision medicine. By identifying these biomarkers, cancer patients may be able to receive treatments they might not otherwise receive.

**GENETICS VS. GENOMICS: WHAT’S THE DIFFERENCE?**

Genetics and genomics might sound similar, but they have different roles in the world of health and disease. Understanding these differences is important to understanding cancer risk, diagnosis, and treatment. **Genetics** is the study of genes and how they determine certain traits passed down from one generation to another. Genetic testing identifies inherited mutations that could increase your risk of developing cancer. These mutations are present in the DNA of your body's cells and can be passed on from one or both parents.

**Genomics** studies a person’s genes and focuses on how your genes interact with each other and with your environment. Acquired or somatic mutations are the most common cause of cancer. Damage to a gene can occur at any time because of environmental factors such as smoking, tobacco use, and radiation. These mutations happen within the tumor itself and are not passed down through generations (not inherited).

**UNDERSTANDING GENETIC COUNSELING: ARE YOU AT RISK?**

Before undergoing genetic testing (which is different from biomarker testing), it's helpful to meet with a genetic counselor. They are experts who can discuss your personal and family medical history related to potential cancer risks. Genetic counselors can guide you through the process of testing and understanding your results with compassion and care.
The presence of a tumor marker alone does not necessarily mean cancer is present or that cancer has reoccurred. These tests are used alongside other diagnostic tools for a comprehensive understanding.

SPECIFIC TESTS TO INFORM CANCER RISK AND TREATMENT

Some of the specific tests that can provide valuable information for cancer risk assessment and treatment decisions include:

1. **Chromosome Testing (also known as Cytogenetic Testing)**: Chromosomes are like tiny instruction manuals in your cells. Sometimes, changes in chromosomes can lead to abnormal proteins, causing cells to grow uncontrollably and become cancerous. Cytogenetic tests help identify these changes and can be used to screen for or diagnose cancer.

2. **Molecular Testing**: This type of testing looks for changes in genes, proteins, or other molecules in tissue samples. For example, lung adenocarcinoma can have a specific mutation in the EGFR gene that leads to abnormal protein production, contributing to the growth and spread of lung cancer.

3. **Biochemical Testing**: When genes mutate, the proteins they produce can become abnormal. Biochemical tests examine these abnormal proteins, which play a role in cancer growth. Immunohistochemistry (IHC) is one form of biochemical testing that measures protein levels in tissue samples. This test can also help determine whether a cancer is responding to treatment. Biochemical testing helps determine treatment responses and is vital in breast cancer when assessing HER2, estrogen receptor (ER), and progesterone receptor (PR) proteins.

4. **Germline Testing**: Your doctor may recommend germline testing based on your family history. Germline testing checks your DNA for genetic mutations present from birth that could increase your risk of developing cancer. Lynch syndrome, for example, is associated with a higher risk of colorectal and endometrial cancers. Knowing if you have an inherited genetic mutation can guide screenings and prevention strategies.

5. **Other Tumor Marker Testing**: Some tumor markers can be used as screening tools, to monitor treatment response, or to detect cancer recurrence. One example is the prostate-specific antigen (PSA) test, which helps screen, diagnose, and monitor prostate cancer.

Ask your medical team if there is a specific test they recommend and how the results will be used in guiding treatment decisions. Contact your insurance plan before testing to see if the specific test being considered is covered and if so, what your cost share will be.

Patient Advocate Foundation can help you address financial or insurance barriers you face with gaining access. Click this link to view our full-length guide entitled *Getting the Right Test at the Right Time: A Cancer Patient’s Guide to Biomarkers*. References for all Biomarker Tip Sheets can be found [here](#).

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