

Need to Know: Key Vocabulary and Terms

Biomarker

A biological molecule found in blood, other body fluids, or tissues that is a sign of a normal or abnormal process, or of a condition or disease. A biomarker may be used to see how well the body responds to a treatment for a disease or condition. Also called molecular marker and signature molecule.

Biomarker testing

A laboratory method that uses a sample of tissue, blood, or other body fluid to check for certain genes, proteins, or other molecules that may be a sign of a disease or condition, such as cancer. Biomarker testing can also be used to check for certain changes in a gene or chromosome that may increase a person's risk of developing cancer or other diseases. Biomarker testing may be done with other procedures, such as biopsies, to help diagnose some types of cancer. It may also be used to help plan treatment, find out how well treatment is working, make a prognosis, or predict whether cancer will come back or spread to other parts of the body. Also called molecular profiling and molecular testing.

Biopsy

The removal of cells or tissues for examination by a pathologist. The pathologist may study the tissue under a microscope or perform other tests on the cells or tissue. There are many different types of biopsy procedures. The most common types include: (1) incisional biopsy, in which only a sample of tissue is removed; (2) excisional biopsy, in which an entire lump or suspicious area is removed; and (3) needle biopsy, in which a sample of tissue or fluid is removed with a needle. When a wide needle is used, the procedure is called a core biopsy. When a thin needle is used, the procedure is called a fine-needle aspiration biopsy.

Clinical trial

A type of research study that tests how well new medical approaches work in people. These studies test new methods of screening, prevention, diagnosis, or treatment of a disease. Also called clinical study.

dMMR

Describes cells that have mutations (changes) in certain genes that are involved in correcting mistakes made when DNA is copied in a cell. dMMR cells usually have many DNA mutations, which may lead to cancer. dMMR is most common in colorectal cancer, other types of gastrointestinal cancer, and endometrial cancer, but it may also be found in cancers of the breast, prostate, bladder, and thyroid and in an inherited disorder called Lynch syndrome. Knowing if a tumor is dMMR may help plan treatment or predict how well the tumor will respond to treatment. Also called deficient DNA mismatch repair, deficient mismatch repair, mismatch repair deficiency, and MMR deficiency.

Driver mutation

A term used to describe changes in the DNA sequence of genes that cause cells to become cancer cells and grow and spread in the body. Checking tumor tissue for driver mutations may help plan treatment to stop cancer cells from growing, including drugs that target a specific mutation.

HER2

A protein involved in normal cell growth. HER2 may be made in larger than normal amounts by some types of cancer cells, including breast, ovarian, bladder, pancreatic, stomach, and esophageal cancers. This may cause cancer cells to grow more quickly and spread to other parts of the body. Checking the amount of HER2 on some types of cancer cells may help plan treatment. Also called c-erbB-2, HER2/neu, human EGF receptor 2, and human epidermal growth factor receptor 2.

Immunohistochemistry

A laboratory method that uses antibodies to check for certain antigens (markers) in a sample of tissue. The antibodies are usually linked to an enzyme or a fluorescent dye. After the antibodies bind to the antigen in the tissue sample, the enzyme or dye is activated, and the antigen can then be seen under a microscope. Immunohistochemistry is used to help diagnose diseases, such as cancer. It may also be used to help tell the difference between different types of cancer.

Immunotherapy

A type of therapy that uses substances to stimulate or suppress the immune system to help the body fight cancer, infection, and other diseases. Some types of immunotherapy only target certain cells of the immune system. Others affect the immune system in a general way. Types of immunotherapy include cytokines, vaccines, bacillus Calmette-Guerin (BCG), and some monoclonal antibodies.

Liquid biopsy

A laboratory test done on a sample of blood, urine, or other body fluid to look for cancer cells from a tumor or small pieces of DNA, RNA, or other molecules released by tumor cells into a person's body fluids. Liquid biopsy allows multiple samples to be taken over time, which may help doctors understand what kind of genetic or molecular changes are taking place in a tumor. A liquid biopsy may be used to help find cancer at an early stage. It may also be used to help plan treatment or to find out how well treatment is working or if cancer has come back.

MSI-high cancer

Describes cancer cells that have a high number of mutations (changes) within microsatellites. For example, microsatellite testing that shows mutations in 30% or more microsatellites is called microsatellite instability-high. Microsatellites are short, repeated sequences of DNA. MSI-H cancer cells may have a defect in the ability to correct mistakes that occur when DNA is copied in the cell. Microsatellite instability is found most often in colorectal cancer, gastric cancer, and

endometrial cancer, but it may also be found in many other types of cancer. Knowing whether cancer is microsatellite instability-high may help plan the best treatment. Also called microsatellite instability-high cancer.

Molecular testing

A laboratory method that uses a sample of tissue, blood, or other body fluid to check for certain genes, proteins, or other molecules that may be a sign of a disease or condition, such as cancer. Molecular testing can also be used to check for certain changes in a gene or chromosome that may increase a person's risk of developing cancer or other diseases. Molecular testing may be done with other procedures, such as biopsies, to help diagnose some types of cancer. It may also be used to help plan treatment, find out how well treatment is working, make a prognosis, or predict whether cancer will come back or spread to other parts of the body. Also called biomarker testing and molecular profiling.

Mutation

Any change in the DNA sequence of a cell. Mutations may be caused by mistakes during cell division, or they may be caused by exposure to DNA-damaging agents in the environment. Mutations can be harmful, beneficial, or have no effect. If they occur in cells that make eggs or sperm, they can be inherited; if mutations occur in other types of cells, they are not inherited. Certain mutations may lead to cancer or other diseases. A mutation is sometimes called a variant.

NTRK gene fusion

A mutation (change) that occurs when a piece of the chromosome containing a gene called NTRK breaks off and joins with a gene on another chromosome. NTRK gene fusions lead to abnormal proteins called TRK fusion proteins, which may cause cancer cells to grow. NTRK gene fusions may be found in some types of cancer, including cancers of the brain, head and neck, thyroid, soft tissue, lung, and colon. Also called neurotrophic tyrosine receptor kinase gene fusion.

PD-L1

A protein that acts as a kind of "brake" to keep the body's immune responses under control. PD-L1 may be found on some normal cells and in higher-than-normal amounts on some types of cancer cells. When PD-L1 binds to another protein called PD-1 (a protein found on T cells), it keeps T cells from killing the PD-L1-containing cells, including the cancer cells. Anticancer drugs called immune checkpoint inhibitors bind to PD-L1 and block its binding to PD-1. This releases the "brakes" on the immune system and leaves T cells free to kill cancer cells.

Personalized medicine

A form of medicine that uses information about a person's own genes or proteins to prevent, diagnose, or treat disease. In cancer, personalized medicine uses specific information about a person's tumor to help make a diagnosis, plan treatment, find out how well treatment is

working, or make a prognosis. Examples of personalized medicine include using targeted therapies to treat specific types of cancer cells, such as HER2-positive breast cancer cells, or using tumor marker testing to help diagnose cancer. Also called precision medicine.

Rare disease

A disease that affects fewer than 200,000 people in the United States. There are about 7,000 rare diseases. An estimated 25 million to 30 million Americans are living with a rare disease. The cause of many rare diseases is unknown, but they are often caused by changes in a person's genes or chromosomes. Rare diseases are often more difficult to diagnose and treat than the more common diseases. Also called rare disorder.

Targeted Therapy

A type of treatment that uses drugs or other substances to target specific molecules that cancer cells need to survive and spread. Targeted therapies work in different ways to treat cancer. Some stop cancer cells from growing by interrupting signals that cause them to grow and divide, stopping signals that help form blood vessels, delivering cell-killing substances to cancer cells, or starving cancer cells of hormones they need to grow. Other targeted therapies help the immune system kill cancer cells or directly cause cancer cell death. Most targeted therapies are either small-molecule drugs or monoclonal antibodies. Also called molecularly targeted therapy.

Tumor mutational burden

The total number of mutations (changes) found in the DNA of cancer cells. Knowing the tumor mutational burden may help plan the best treatment. For example, tumors that have a high number of mutations appear to be more likely to respond to certain types of immunotherapy. Tumor mutational burden is being used as a type of biomarker. Also called TMB.

*These terms originally appeared in the [National Cancer Institute Dictionary of Cancer Terms](#).