Genetic and Biomarker Testing for Cancer

Talking with your doctor or care team about genetic or biomarker testing can involve words you might not know or understand. Organizations like Cancer.net, the National Cancer Institute and the American Cancer Society are great resources for looking up definitions for cancer terms your care team may use.

Below, we’ve compiled a list of common words that may come up when discussing genetic and biomarker testing and how it can help with diagnosis or determine appropriate cancer treatment with your care team. Remember to always ask your doctor to explain any words you don’t know, so that you can understand your cancer care plan.

1 GENES
Genes contain the instructions needed for cells to do their specific job. Genes are made up of deoxyribonucleic acid (DNA). The building blocks of DNA are arranged in a specific sequence that defines each gene.

2 MUTATION
A mutation is a change in a gene that impacts how the gene functions. Some mutations are inherited, meaning they are present at birth and get passed down from parents to their children. Other mutations are acquired, meaning they develop later in life and are not passed down to the next generation. Mutations can lead to many types of disease, including cancer.

3 PROTEINS
Proteins perform specific functions within a cell. For a cell to work normally, genes must provide the right instructions for making its proteins.

4 GENETIC TESTING
Genetic testing looks for inherited mutations in a person’s genes and may be used to figure out the risk of developing cancer.

5 DNA SEQUENCING
A method used to determine the exact order of the building blocks that make up DNA. This method can be used to find changes in the DNA that may cause cancer.

6 GENOMIC SEQUENCING
A method used to determine the entire genetic makeup of a person. This method can be used to find changes in areas of the genetic makeup to help scientists understand how certain cancers form. Results of genomic sequencing may be used to diagnose and treat cancer.

7 BIOMARKER TESTING
A biomarker test analyzes a person’s biological information, such as their cells or genes. This can be done by looking at substances in your blood, tissues or other body fluids that can be used to help learn more about your cancer. These tests may help you and your doctor select a cancer treatment for you if a certain biomarker is present.

8 BIOPSY
A procedure performed by a doctor where cells or tissue is removed from the body and looked at for signs of disease, such as cancer. If there is evidence of cancer, the biopsy results can show what type of cancer it is.

9 PRECISION MEDICINE
An approach to health care where specific plans are based on a person’s genes. In cancer care, precision medicine looks at how a specific gene change (gene mutation) might affect a person’s risk of getting a certain cancer or, if they already have cancer, how their genes might affect treatment.

10 TARGETED CANCER THERAPY
A type of treatment that attacks specific parts of a cell, like proteins, thought to be involved in the development, growth and spread of cancer.