

Key Terms on Biomarker Testing

A **biomarker**, or biological marker, is a characteristic of the body that is a sign of a normal or abnormal process, condition, or disease. Biomarkers can be used in the diagnosis, prognosis, monitoring, and/or treatment selection of different conditions and diseases, such as brain and spinal tumors.

Biomarker testing, also called molecular testing, somatic testing, and tumor profiling, is a laboratory test used to analyze tissue, blood, or other bodily fluids for specific mutations, gene alterations, proteins, and/or other biomarkers. Biomarker testing of brain and spinal tumors can help individuals and their care teams identify the correct diagnosis, prognosis, and appropriate treatment options. Biomarker testing can involve single analyte tests, which examine a single biomarker; multiple-analyte panels, which examine a set of biomarkers; and more complex tests, such as next-generation sequencing (NGS).

A **biopsy** is a medical procedure to remove a sample of tissue or cells so that it can be examined by a pathologist. The pathologist may study the sample under a microscope and/or perform other tests, such as biomarker testing.

A **biospecimen** is a sample of tissue, blood, or other bodily fluid.

A **gene** is the basic unit of inheritance. Genes are pieces of DNA passed from parents to children that contain the information needed to specify physical and biological traits.

Genetic testing is the use of a laboratory test to examine an individual's DNA. In the medical setting, genetic testing may be used to determine a person's risk of developing a disease or condition or having a child with the disease or condition; this type of genetic testing is considered genetic testing for inherited risk. Genetic testing performed on tumor tissue that helps diagnose, prognose, and plan treatment is called biomarker testing.

Genetic testing for inherited cancer risk, also called germline testing, is a laboratory test that can show if an individual has inherited a genetic change that increases their risk of cancer. While cancer itself cannot be passed down from parents to children, a genetic change that increases the risk of cancer can be passed down if it is present in the parent's egg or sperm cells.

A tumor grade is a number used to describe how abnormal the tumor cells and tissue look under

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a microscope when compared to healthy cells and tissue. Grades can be 1, 2, 3, or 4, with lower numbers considered "low-grade" tumors and higher numbers considered "high-grade" tumors. Low-grade tumors tend to appear more like healthy cells and tissue under a microscope. High-grade tumors tend to appear more abnormal and disorganized under a microscope.

Histopathology is the study of tissue or cells under a microscope to look for the presence of disease.

A **mutation** is a change in one's DNA that can result from a random mistake or exposure to a substance, organism, or agent capable of inducing changes in DNA. Mutations can cause a harmful, beneficial, or neutral effect on the individual. Mutations that occur in germ cells like egg and sperm cells are called germline mutations and can be passed on to one's children. Mutations that occur in other cells of the body are called somatic mutations and are not passed on.

Next-generation sequencing, abbreviated NGS, is a newer technology used to sequence DNA and detect mutations that is faster and less expensive than the earlier, first-generation method. For people with brain and spinal tumors, next-generation sequencing can help care teams identify the diagnosis, prognosis, and treatment options.

Precision medicine, also called personalized medicine or individualized medicine, is an approach that uses information about an individual's genes, environment, and lifestyle to guide decisions related to their medical management. Biomarker testing can help care teams determine a more precise diagnosis and determine targeted treatment options for people with brain and spinal tumors.

The WHO Classification of Tumours, also known as the WHO Blue Book, provides an evidence-based classification system of tumors to standardize diagnosis and improve patient care worldwide. The 2021 WHO Classification of Tumors of the Central Nervous System, the current gold standard for brain and spinal tumors, emphasizes the importance of an integrated diagnosis, which layers information about the tumor's histopathology, grade, and biomarkers.

References

Duffy M. J. (2001). Clinical uses of tumor markers: a critical review. Critical reviews in clinical laboratory sciences, 38(3), 225–262. https://doi.org/10.1080/20014091084218

Duffy M. J. (2013). Tumor markers in clinical practice: a review focusing on common solid cancers. Medical principles and practice: international journal of the Kuwait University, Health Science Centre, 22(1), 4–11. https://doi.org/10.1159/000338393

Louis, D. N., Perry, A., Wesseling, P., Brat, D. J., Cree, I. A., Figarella-Branger, D., Hawkins, C., Ng, H. K., Pfister, S. M.,

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Reifenberger, G., Soffietti, R., von Deimling, A., & Ellison, D. W. (2021). The 2021 WHO classification of tumors of the central nervous system: a summary. Neuro-oncology, 23(8), 1231–1251. https://doi.org/10.1093/neuonc/noab106

Martin, N. A., Tepper, J. E., Giri, V. N., Stinchcombe, T. E., Cheng, H. H., Javle, M. M., & Konnick, E. Q. (2021). Adopting consensus terms for testing in precision medicine. JCO precision oncology, 5, PO.21.00027. https://doi.org/10.1200/PO.21.00027

National Academies of Sciences, Engineering, and Medicine. (2016). Biomarker tests for molecularly targeted therapies: Key to unlocking precision medicine. National Academies Press. https://doi.org/10.17226/21860

National Cancer Institute. (n.d.). NCI dictionary of cancer terms. Retrieved February 15, 2024, from https://www.cancer.gov/publications/dictionaries/cancer-terms

National Cancer Institute. (2021, December 14). Biomarker testing for cancer treatment. Retrieved February 15, 2024, from https://www.cancer.gov/about-cancer/treatment/types/biomarker-testing-cancer-treatment

National Human Genome Research Institute. (n.d.). Talking glossary of genomic and genetic terms. Retrieved February 15, 2025, from https://www.genome.gov/genetics-glossary

PDQ® Cancer Genetics Editorial Board. (2024, February 2). Cancer genetics overview (PDQ®). National Cancer Institute. Retrieved February 15, 2024, from https://www.cancer.gov/about-cancer/causes-prevention/genetics/overview-pdq

PDQ[®] Cancer Genetics Editorial Board. (2024, February 2). Cancer genetics risk assessment and counseling (PDQ[®]). National Cancer Institute. Retrieved February 15, 2024, from https://www.cancer.gov/about-cancer/causes-prevention/genetics/risk-assessment-pdq

Qin D. (2019). Next-generation sequencing and its clinical application. Cancer biology & medicine, 16(1), 4–10. https://doi. org/10.20892/j.issn.2095-3941.2018.0055

WHO Classification of Tumours Editorial Board. (2021). Central nervous system tumours (5th ed., Vol. 6, Ser. WHO classification of tumours series). International Agency for Research on Cancer. https://publications.iarc.fr/601