

Laboratory Medicine in the Era of Disruptive Technology

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## Tumor markers- laboratory tests used in cancer diagnosis

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Cancer is the name given to a collection of heterogeneous but related diseases that can start in most parts of the body and that are characterized by uncontrolled division of cells that ultimately may spread into surrounding tissues. Early detection of malignancy optimizes any opportunities for curative surgery for some in situ cancers. Most cancers do not produce symptoms until tumors are too large to be removed surgically or until cancerous cells have spread to other tissue either by invading local lymph nodes or by distant spread (metastasis) to other organs. Systemic treatments (chemotherapy, endocrine therapy, or immunotherapy) are then usually the only options but are not curative. Any residual viable cancerous cells remaining after treatment may proliferate, develop resistance to further therapy, and ultimately cause the death of the patient.

Tumor markers are substances that may be present in abnormally high concentrations in body fluids or tissue from patients with cancer. Tumor markers can aid cancer management in a number of ways, including screening, diagnosis, prognostic assessment, therapy prediction, and/or posttreatment monitoring. Ideal tumor marker includes the following: 1. Detectable only in a given malignancy and absent in the healthy population or in nonmalignant conditions (ie, with clinical specificity and clinical sensitivity approaching 100%). 2. Present in a readily acquired biological matrix (eg, serum, urine, tumor tissue). 3. Present at concentrations proportional to tumor burden. 4. Conveniently measured by a readily available, simple, reproducible, and inexpensive procedure. 5. Beneficial to clinical care with a measurable effect on patient outcome. No such tumor marker has yet been identified.

Reflecting the heterogeneous nature of cancer, tumor markers encompass a variety of molecular species, which may be tumor-derived or tumor-associated. Many tumor markers are produced by a variety of different tumors, and few tumor markers are organ-specific or specific for a single type of malignancy. Tumor markers range from simple molecules (eg, catecholamines) to relatively well-characterized proteins (eg, hormones, enzymes, and gene products) to much more heterogeneous glycoproteins and mucins (eg, CA125), which may be defined by the antibodies used to measure them. Several important tumor markers (eg, AFP, CEA, and hCG) are oncofetal antigens, which are present in the fetus in normal pregnancy but are expressed at high concentrations in tissue or body fluids of adults with some cancers. Tumor markers are present in cells, tissues, or body fluids and can be measured either qualitatively or quantitatively using chemical, immunologic, or molecular biological methods. While their structures and properties vary widely, the broad principles underpinning their evidence-based clinical application are common to all tumor markers. Tumor markers are surrogate indicators that can help to increase or decrease the clinical suspicion that a future clinically important event, such as the development of a new or secondary cancer, recurrence, progression, or death will or will not occur, and/or that a specific treatment will reduce that risk.

Tumor marker measurements contribute significantly to the management of cancer patients, but considerable care is required to ensure that their use is appropriate. The clinical laboratory should proactively encourage correct test selection, sample handling, analysis, reporting, and interpretation of tumor marker results, taking particular care to remind users of the caveats associated with these tests. This is essential not only for the well-established tumor markers but also for the new generation





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of molecular and genetic tumor markers on which clinicians increasingly will rely to provide optimal care.

Reference

1. Tietz textbook of clinical chemistry and molecular diagnostics, 6<sup>th</sup> Edition. 2017:436-78.

