



LABORATORY DIAGNOSTICS OF ONCOLOGICAL DISEASES: ADVANCES AND CLINICAL APPLICATIONS

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Annotation

Oncological diseases, or cancers, represent one of the leading causes of morbidity and mortality worldwide. Over the years, advances in laboratory diagnostics have significantly improved the early detection, monitoring, and treatment of cancer. The role of laboratory tests in oncology is pivotal, as they provide essential information on tumor biomarkers, genetic mutations, tumor profiling, and response to therapies. This article aims to explore the current state of laboratory diagnostics in oncology, emphasizing the various diagnostic techniques, biomarkers, and future directions in cancer diagnostics.

Keywords: cancer, laboratory diagnostics, biomarkers, diagnosis.

Introduction

Cancer arises from the uncontrolled growth of abnormal cells, often due to genetic mutations and alterations in cellular processes. Early detection plays a critical role in improving survival rates, and laboratory diagnostics are at the forefront of identifying cancer-related biomarkers, detecting the presence of tumors, and monitoring treatment effectiveness. Over the past few decades, scientific



International Conference on Educational Discoveries and Humanities

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16th April, 2025

advancements have led to the development of more accurate and efficient laboratory tests that have greatly impacted the clinical management of oncological diseases [4,6].

Laboratory diagnostics in oncology are vital for several key purposes:

- Early detection: detecting cancer in its early stages significantly improves the chances of successful treatment;
- Diagnosis confirmation: laboratory tests, in combination with imaging and clinical examination, help confirm the diagnosis of cancer;
- Prognosis prediction: certain biomarkers can provide insight into the aggressiveness of cancer, its potential for metastasis, and overall prognosis;
- Treatment monitoring: monitoring the effectiveness of therapies, detecting relapse, and adjusting treatment plans;
- Personalized medicine: genetic profiling of tumors to guide the selection of targeted therapies and improve patient outcomes.

Several laboratory techniques are used to diagnose, monitor, and guide the treatment of oncological diseases [2,3,5]. These techniques include blood tests, tissue biopsies, molecular diagnostics, and genetic testing.

1. Biopsy and histopathology.

Tissue biopsy remains the gold standard for cancer diagnosis. A sample of tissue is removed from the tumor and examined under a microscope to assess cellular morphology, identify tumor types, and determine malignancy. Histopathological examination allows for: tumor grading, tumor staging, histological subtyping, identifying the specific type of cancer, such as adenocarcinoma, squamous cell carcinoma, or lymphoma.

2. Blood-based biomarkers.

Blood tests are essential for detecting various biomarkers that can suggest the presence of cancer. Blood-based biomarkers are proteins, genes, or molecules released by cancer cells into the bloodstream. Common blood-based biomarkers include:



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- Prostate- specific antigen (PSA): Used for the early detection of prostate cancer. Elevated PSA levels may indicate prostate cancer, but they can also be elevated in benign conditions such as prostatitis or benign prostatic hyperplasia (BPH);
- Carcinoembryonic antigen (CEA): Used primarily for colorectal cancer but can also be elevated in cancers of the breast, pancreas, lung, and liver;
- Alpha-fetoprotein (AFP): Elevated in hepatocellular carcinoma (liver cancer) and germ cell tumors;
- Cancer antigen 125 (CA-125): Primarily used for the monitoring of ovarian cancer, although it can be elevated in other conditions, such as endometriosis or pelvic inflammatory disease;
- Cancer antigen 19-9 (CA 19-9): Used for pancreatic, colorectal, and gastric cancers.

These biomarkers are not necessarily diagnostic on their own but are used alongside imaging and clinical evaluation to confirm cancer and monitor disease progression or response to treatment [1,2,4].

3. Molecular diagnostics and genetic testing.

Molecular diagnostics have revolutionized the field of oncology by providing insights into the genetic mutations and alterations that drive cancer development. Genetic testing enables the identification of mutations that may be targeted with specific therapies. Key technologies include:

- Next-generation sequencing (NGS): NGS allows for the sequencing of entire genomes or specific genes to identify mutations, copy number variations, and structural alterations in cancer cells. This technique enables the discovery of genetic abnormalities like EGFR mutations in non-small cell lung cancer (NSCLC) or BRCA1/2 mutations in breast and ovarian cancers;
- Polymerase chain reaction (PCR): PCR amplifies specific genetic sequences to identify mutations or the presence of viral DNA (e.g., Epstein-Barr virus, human papillomavirus). PCR is often used to detect BCR-ABL fusion gene in chronic myelogenous leukemia (CML);



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- Fluorescence in situ hybridization (FISH): A technique used to detect specific chromosomal abnormalities in cancer cells, such as HER2 amplification in breast cancer, which has important therapeutic implications for HER2-targeted therapies;
- Circulating tumor DNA (ctDNA) and liquid biopsies: ctDNA is fragmented DNA that is shed from tumors into the bloodstream. Liquid biopsy techniques detect ctDNA to identify mutations or alterations in genes such as KRAS, BRAF, or PIK3CA. Liquid biopsies are particularly valuable for detecting minimal residual disease or monitoring for relapse in cancers such as lung and colorectal cancer [3,6].

4. Immunohistochemistry (IHC).

Immunohistochemistry (IHC) involves using antibodies to detect specific antigens in tissue samples. IHC is widely used to identify tumor-specific markers and help classify cancers. For example:

- HER2 expression: In breast cancer, HER2 overexpression is associated with aggressive disease, and testing for HER2 helps determine whether the patient will benefit from HER2-targeted therapies such as trastuzumab;
- PD-L1 expression: In cancers like non-small cell lung cancer (NSCLC), testing for PD-L1 expression helps to determine the eligibility for immune checkpoint inhibitors.
- IHC is also used to classify lymphoma and other cancers by detecting the presence of specific tumor markers.

Conclusion

Laboratory diagnostics play an indispensable role in the diagnosis, prognosis, and treatment of oncological diseases. From traditional techniques like biopsy and histopathology to cutting-edge technologies such as liquid biopsies and next-generation sequencing, laboratory tests provide critical insights into cancer biology, enabling earlier detection, personalized therapy, and better clinical outcomes. As technology continues to evolve, the future of cancer diagnostics will increasingly rely on more precise, non-invasive, and cost-effective methods to detect cancer earlier and guide more effective treatments. The combination of molecular



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diagnostics, biomarkers, and artificial intelligence promises to transform cancer care and improve survival rates globally.

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