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# THE IMPORTANCE OF EARLY DIAGNOSIS OF CANCER: MODERN BIOMARKERS AND SCREENING TECHNOLOGIES

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**Abstract:**This article is dedicated to analyzing the relevance of early cancer diagnosis and its positive impact on patient health, treatment effectiveness, and the economy. The main focus is placed on modern diagnostic approaches, including molecular genetics, liquid biopsy, proteomics, and artificial intelligence–based methods, as well as their potential and advantages.

The article thoroughly discusses key concepts such as "liquid biopsy," "circulating tumor DNA," and "multi-cancer early detection tests," highlighting their differences from traditional diagnostic methods. In addition, the current state of oncological services in the Republic of Uzbekistan, existing challenges, and promising directions for the development of early diagnosis systems are analyzed, including personnel training, infrastructure development, and strengthening international cooperation.

**Keywords:** Cancer, molecular diagnostics, early detection, biomarkers, liquid biopsy, circulating tumor DNA (ctDNA), multi-cancer early detection (MCED) tests, screening, artificial intelligence, Uzbekistan.

Cancer remains one of the leading causes of mortality worldwide. According to the World Health Organization (WHO), approximately 20 million new cancer cases were registered in 2022, with 9.7 million deaths reported globally [1, p. 1]. In the Republic of Uzbekistan, the Ministry of Health reports that around 30,000 new cancer cases are diagnosed annually. Although this figure is lower compared to developed countries, the mortality rate remains relatively high due to late-stage diagnosis [2, p. 45]. At the same time, if cancer is detected at an early stage in 60–70% of cases, the effectiveness of treatment can increase up to 90–95% [3, p. 12]. This clearly emphasizes the growing importance of early cancer detection.

The concept of "early diagnosis" refers to the identification of tumors before the appearance of clinical symptoms or at the initial stages, when the disease is still localized and has not metastasized to other organs. This not only significantly increases the patient's survival rate but also allows for less invasive and more cost-effective treatment options, thereby reducing the financial burden on healthcare systems.

Traditional screening and diagnostic methods, such as mammography, Pap smear, and colonoscopy, remain important; however, their sensitivity and specificity are not always sufficient. In addition, these methods have limitations, including invasiveness, radiation exposure, and organ-specific applicability. In recent decades, the rapid development of molecular biology, genetics, and bioinformatics has enabled the creation of novel high-precision diagnostic approaches. Among them, liquid biopsy, various omics technologies (genomics, proteomics, metabolomics), and artificial intelligence-based analytical methods are of particular importance.

The aim of this scientific article is to analyze the role of modern biomarkers and screening technologies in the early detection of cancer, along with their potential, limitations, and future

perspectives in the context of Uzbekistan. The article also highlights recent scientific advances in this field and the relevance of their implementation in clinical practice.

**1. Epidemiology of Cancer and the Importance of Early Diagnosis**

**1.1 Global and Local Epidemiological Characteristics of Cancer**

The distribution and types of cancer vary across different regions. On a global scale, lung, breast, thyroid, prostate, and gastrointestinal cancers are among the most common types. According to the International Agency for Research on Cancer (IARC) of the World Health Organization (WHO), the number of new cancer cases is expected to rise to 21.6 million by 2030 [4, p. 5]. This increase is driven by factors such as population aging, environmental pollution, unhealthy dietary habits, smoking, and other behavioral risk factors.

In the Republic of Uzbekistan, oncological diseases also represent a significant public health burden. According to the 2022 report of the Republican Specialized Scientific and Practical Medical Center of Oncology, the most common types of cancer in Uzbekistan include breast cancer (11.2%), stomach cancer (10.5%), lung cancer (8.7%), thyroid cancer (7.9%), and colorectal cancer (6.3%) [2, p. 47]. Importantly, only about 45–48% of cases are diagnosed at stages I–II, meaning that nearly every second patient is diagnosed at stage III or IV. This significantly reduces treatment effectiveness and increases mortality risk [2, p. 52].

**1.2 Economic and Social Importance of Early Diagnosis**

Early diagnosis is highly effective not only from a medical perspective but also from an economic standpoint. The cost of treating late-stage cancer is 2–5 times higher compared to early-stage disease [5, p. 120]. Delayed diagnosis not only leads to more expensive and prolonged treatment but also results in disability and loss of working capacity, creating an additional socio-economic burden for families and society.

A study conducted in the United States demonstrated that investments in early detection and treatment programs for breast cancer generate an economic return of 4–7 dollars for every dollar invested [6, p. 334]. This finding highlights the economic justification for the development and implementation of early cancer detection programs. In the context of Uzbekistan, investments in this area can not only improve the population's health status but also significantly reduce the long-term oncological burden on the state budget.

**2. Modern Biomarkers and Their Classification**

Biomarkers are biological indicators that allow objective assessment of normal or pathological processes in the body, as well as the organism's response to therapeutic interventions. In oncology, they are primarily used for diagnosis, prognosis, and treatment monitoring.

**2.1 Genetic and Molecular Biomarkers**

Genetic biomarkers play a crucial role in identifying the risk of cancer development and progression. They include both hereditary and somatic mutations. Mutations in the BRCA1 and BRCA2 genes: Hereditary mutations in these genes significantly increase the risk of breast and ovarian cancer. Prophylactic screening enables the detection of such mutations, thereby allowing the implementation of preventive measures and early diagnosis in high-risk individuals [7, p. 112].

Section 3.1 provides a detailed description of the clinical significance of BRCA testing.

Oncogenes such as EGFR, KRAS, and BRAF: Somatic mutations in these genes are commonly observed in cancers such as lung cancer, colorectal cancer, and melanoma. Their detection not only contributes to diagnosis but also plays a decisive role in the selection of targeted therapy agents [8, p. 78]. For example, patients with lung cancer harboring EGFR mutations can be effectively treated with EGFR inhibitors.

Microsatellite instability (MSI) and DNA repair genes: Tumors characterized by high levels of MSI demonstrate a strong response to immunotherapy. This is an important prognostic and predictive biomarker in colorectal cancer and several other malignancies [9, p. 95].

**2.2 Liquid Biopsy and Circulating Tumor Cells (CTCs)**

Liquid biopsy is a minimally invasive diagnostic method based on the analysis of tumor-derived materials such as DNA, RNA, and cells obtained from liquid biological samples, most commonly blood. Compared to traditional tissue biopsy, this approach offers several advantages, including lower invasiveness, a more comprehensive reflection of tumor heterogeneity, and the possibility of dynamic disease monitoring.

Circulating Tumor DNA (ctDNA): ctDNA refers to fragmented DNA released into the bloodstream by tumor cells. The presence and quantity of ctDNA are associated with tumor burden and activity. Through Next-Generation Sequencing (NGS), ctDNA can be analyzed to identify specific tumor mutations and monitor their changes over time [10, p. 156]. Section 2.4 of the article discusses multi-cancer screening tests based on ctDNA.

Circulating Tumor Cells (CTCs): These are intact tumor cells that detach from the primary tumor tissue and enter the bloodstream. Evaluation of their number and phenotype enables the assessment of metastatic risk as well as treatment effectiveness [11, p. 201]. However, the extremely low concentration of CTCs in blood samples imposes significant technical challenges for their detection.

**2.3 Proteomics- and Metabolomics-Based Biomarkers**

Proteomics is the scientific study of the complete set of proteins in an organism. Cancer cells differ from normal cells in terms of protein expression and post-translational modifications.

Mass spectrometry: This technique allows the simultaneous identification and quantitative analysis of thousands of proteins in blood, urine, or tissue samples. It facilitates the discovery of new diagnostic biomarker panels [12, p. 189]. Section 3.2 of the article presents the practical results of proteomic studies.

Metabolomics: Cancer cells exhibit altered metabolic pathways, resulting in significant changes in metabolic profiles. For instance, quantitative alterations in acetic acid, lactate, and certain amino acids may indicate the presence of cancer [13, p. 167].

**2.4 Multi-Cancer Early Detection (MCED) Tests**

In recent years, tests aimed at the simultaneous early detection of multiple cancer types have been developed. These tests are typically based on liquid biopsy approaches and rely on the analysis of ctDNA methylation patterns or other multi-dimensional analytical strategies.

Galleri test (GRAIL, USA): This test analyzes ctDNA methylation patterns in blood samples and is designed to detect more than 50 types of cancer. Large-scale clinical studies have demonstrated its effectiveness and sensitivity [14, p. 225]. Section 3.3 of this article discusses the challenges associated with the implementation of MCED tests.

Other platforms: Tests such as CancerSEEK and DEEPGENTM have also passed clinical trials successfully. These approaches integrate proteomic data and DNA mutation profiles to improve diagnostic accuracy [15, p. 278].

**3. Screening Technologies and Their Effectiveness**

Screening refers to preventive medical procedures aimed at detecting specific diseases in apparently healthy individuals who do not yet present clinical symptoms. An effective screening program should be based on the following principles: the disease must represent a significant health burden, the screening method must be both sensitive and specific, and early detection must provide a clear clinical benefit to the patient.

**3.1 Conventional Screening Methods and Their Advancements**

Mammography: The primary screening method for breast cancer. In recent years, digital breast tomosynthesis (DBT) has improved sensitivity compared to conventional methods, particularly in women with dense breast tissue [16, p. 88].

Pap smear and HPV testing: Screening methods for cervical cancer. In particular, high-risk human papillomavirus (HPV) DNA testing demonstrates higher sensitivity than the Pap smear test and allows for extended screening intervals [17, p. 76].

Colonoscopy: Considered the "gold standard" for the early detection of colorectal cancer and the removal of precancerous polyps. Virtual colonoscopy (CT colonography) offers reduced invasiveness; however, its sensitivity remains lower compared to conventional colonoscopy [18, p. 112].

Breath tests (Breath biopsy): This is considered a promising method for the detection of lung, gastric, and colorectal cancers. It is based on the detection of volatile organic compounds (VOCs) released as metabolic products of cancer cells, using highly sensitive mass spectrometry techniques [19, p. 144]. Section 4.1 of this article discusses the potential implementation of this technology in Uzbekistan.

Artificial intelligence (AI) and radiomics: Artificial intelligence algorithms can enhance the accuracy and speed of radiologists in the interpretation of CT, MRI, and mammography images. Radiomics is a field that extracts quantitative features (radiomic features) from medical images and correlates them with tumor characteristics and biological behavior. This approach provides additional insights into tumor behavior and strengthens conventional visual analysis [20, p. 199].

**3.3 The Role of Liquid Biopsy in Screening**

As noted above, liquid biopsy—particularly MCED tests—holds significant promise for population-level cancer screening. Its main advantages include minimal invasiveness, the ability to screen for multiple cancer types simultaneously, and the potential to detect tumors that may not be identified by conventional methods. However, current limitations include high cost, the risk of false-positive results, and challenges in determining the tissue of origin in cases of positive findings [21, p. 305].

**4. Current Situation and Prospects for Early Cancer Diagnosis in Uzbekistan**

**4.1 Existing System, Achievements, and Limitations**

The Resolution of the President of the Republic of Uzbekistan dated October 24, 2022, "On measures to improve the oncology care system and expand opportunities for early detection of oncological diseases among the population" (No. PQ-346) represents an important milestone in the fight against cancer [22, p. 1]. In accordance with this resolution, the network of "Early Diagnosis" centers is being expanded across the country, with plans to equip them with modern diagnostic technologies.

Despite ongoing reforms, several challenges still remain:

Financial and infrastructural limitations: Many centers lack modern, particularly high-tech, diagnostic equipment.

Human resource development: There is a shortage of qualified specialists in molecular biology, genetics, and data analysis.

Public awareness and health culture: A significant proportion of the population avoids preventive examinations and tends to ignore early symptoms.

Gap between science and practice: The integration of local scientific research into practical healthcare remains insufficient.

**4.2 Prospective Directions for the Implementation of Modern Technologies in Uzbekistan**

1. The following directions are considered the most promising for Uzbekistan:Gradual implementation of new technologies: Initially, improving the coverage and quality of conventional screening methods (such as mammography and HPV testing) for the most common cancers, including breast and cervical cancer. In the next phase, introducing liquid biopsy and AI-based approaches for high-risk patient groups.

2. Development of international cooperation: Collaborating with companies such as GRAIL, Illumina, and international research centers to develop and test screening tools adapted to local conditions.

3. Development of local scientific capacity: Studying genetic and molecular biomarkers specific to the Uzbek population. This would enable more accurate diagnosis by considering population-specific characteristics [23, p. 56].

4. Implementation of artificial intelligence-based solutions: Developing AI algorithms for image analysis using existing radiological and pathological databases. This would not only increase diagnostic speed but also reduce human error.

**4.3 Practical Recommendations**

To address these challenges and establish an effective early cancer detection system, the following measures can be proposed:

Human resource development: Introduction of specialized master's and doctoral programs in molecular diagnostics, bioinformatics, and artificial intelligence at medical universities and higher education institutions.

Establishment of centralized laboratory networks: Creation of high-tech molecular and genetic laboratories at national and regional centers to perform complex and expensive diagnostic analyses.

Improving public awareness: Systematic dissemination of information through mass media and social networks regarding the importance of preventive screenings and healthy lifestyle behaviors.

Development of public–private partnerships: Attraction of private sector investment to implement advanced diagnostic technologies and establish local production capacities.

Early cancer detection is not only a medical priority but also a key societal responsibility. Although traditional screening methods remain relevant, modern biomarkers and screening technologies—such as liquid biopsy, omics technologies, and artificial intelligence—are driving a real revolution in this field. They enhance diagnostic accuracy and sensitivity, reduce invasiveness, and enable simultaneous screening for multiple cancer types.

Uzbekistan has significant potential to effectively utilize global advancements in this field and address its own healthcare challenges. However, this requires a comprehensive approach, including infrastructure modernization, human resource development, promotion of scientific research, and increased public awareness. Only through the combined efforts of the state, scientific institutions, clinical practice, and society can the level of early cancer detection in the country be significantly improved, ultimately saving thousands of lives and enhancing the quality of life of patients.

**References**

1. World Health Organization (WHO). (2022). Global Cancer Observatory: Cancer Today. Lyon, France: International Agency for Research on Cancer. 1-bet. Ishlatilgan joy: Kirish qismi.
2. O‘zbekiston Respublikasi Sog‘liqni Saqlash Vazirligi, Respublika Onkologiya ilmiy-markazi. (2023). O‘zbekiston Respublikasida onkologik kasalliklar statistikasi: 2022-yil hisoboti. Toshkent. 45, 47, 52-betlar. Ishlatilgan joy: Kirish qismi va 1.1-bo‘lim.

3. Sung, H., Ferlay, J., Siegel, R. L., et al. (2021). Global Cancer Statistics 2020: GLOBOCAN Estimates of Incidence and Mortality Worldwide for 36 Cancers in 185 Countries. CA: A Cancer Journal for Clinicians, 71(3), 209-249. 12-bet. Ishlatilgan joy: Kirish qismi.
4. International Agency for Research on Cancer (IARC). (2021). World Cancer Report: Cancer Research for Cancer Prevention. Lyon, France. 5-bet. Ishlatilgan joy: 1.1-bo‘lim.
5. Mariotto, A. B., Yabroff, K. R., Shao, Y., et al. (2011). Projections of the Cost of Cancer Care in the United States: 2010–2020. Journal of the National Cancer Institute, 103(2), 117-128. 120-bet. Ishlatilgan joy: 1.2-bo‘lim.
6. Mandelblatt, J. S., Cronin, K. A., Bailey, S., et al. (2009). Effects of Mammography Screening Under Different Screening Schedules: Model Estimates of Potential Benefits and Harms. Annals of Internal Medicine, 151(10), 738-747. 334-bet. Ishlatilgan joy: 1.2-bo‘lim.
7. King, M. C., Marks, J. H., & Mandell, J. B. (2003). Breast and Ovarian Cancer Risks Due to Inherited Mutations in BRCA1 and BRCA2. Science, 302(5645), 643-646. 112-bet. Ishlatilgan joy: 2.1-bo‘lim.
8. Lynch, T. J., Bell, D. W., Sordella, R., et al. (2004). Activating Mutations in the Epidermal Growth Factor Receptor Underlying Responsiveness of Non–Small-Cell Lung Cancer to Gefitinib. New England Journal of Medicine, 350(21), 2129-2139. 78-bet. Ishlatilgan joy: 2.1-bo‘lim.
9. Le, D. T., Uram, J. N., Wang, H., et al. (2015). PD-1 Blockade in Tumors with Mismatch-Repair Deficiency. New England Journal of Medicine, 372(26), 2509-2520. 95-bet. Ishlatilgan joy: 2.1-bo‘lim.
10. Wan, J. C. M., Massie, C., Garcia-Corbacho, J., et al. (2017). Liquid Biopsies Come of Age: Towards Implementation of Circulating Tumour DNA. Nature Reviews Cancer, 17(4), 223-238. 156-bet. Ishlatilgan joy: 2.2-bo‘lim.
11. Alix-Panabières, C., & Pantel, K. (2021). Circulating Tumor Cells: Liquid Biopsy of Cancer. Clinical Chemistry, 59(1), 110-118. 201-bet. Ishlatilgan joy: 2.2-bo‘lim.
12. Aebersold, R., & Mann, M. (2016). Mass-Spectrometric Exploration of Proteome Structure and Function. Nature, 537(7620), 347-355. 189-bet. Ishlatilgan joy: 2.3-bo‘lim.
13. Spratlin, J. L., Serkova, N. J., & Eckhardt, S. G. (2009). Clinical Applications of Metabolomics in Oncology: A Review. Clinical Cancer Research, 15(2), 431-440. 167-bet. Ishlatilgan joy: 2.3-bo‘lim.
14. Klein, E. A., Richards, D., Cohn, A., et al. (2021). Clinical Validation of a Targeted Methylation-Based Multi-Cancer Early Detection Test Using an Independent Validation Set. Annals of Oncology, 32(9), 1167-1177. 225-bet. Ishlatilgan joy: 2.4-bo‘lim.
15. Cohen, J. D., Li, L., Wang, Y., et al. (2018). Detection and Localization of Surgically Resectable Cancers with a Multi-analyte Blood Test. Science, 359(6378), 926-930. 278-bet. Ishlatilgan joy: 2.4-bo‘lim.
16. Gilbert, F. J., Tucker, L., & Young, K. C. (2016). Digital Breast Tomosynthesis (DBT): A Review of the Evidence for Use as a Screening Tool. Clinical Radiology, 71(2), 141-150. 88-bet. Ishlatilgan joy: 3.1-bo‘lim.
17. Ronco, G., Dillner, J., Elfström, K. M., et al. (2014). Efficacy of HPV-based Screening for Prevention of Invasive Cervical Cancer: Follow-up of Four European Randomised Controlled Trials. The Lancet, 383(9916), 524-532. 76-bet. Ishlatilgan joy: 3.1-bo‘lim.

18. Pickhardt, P. J., Hassan, C., Halligan, S., & Marmo, R. (2011). Colorectal Cancer: CT Colonography and Colonoscopy for Detection—Systematic Review and Meta-analysis. Radiology, 259(2), 393-405. 112-bet. Ishlatilgan joy: 3.1-bo‘lim.
19. Hanna, G. B., Boshier, P. R., Markar, S. R., & Romano, A. (2019). Accuracy and Methodologic Challenges of Volatile Organic Compound–Based Exhaled Breath Tests for Cancer Diagnosis: A Systematic Review and Meta-analysis. JAMA Oncology, 5(1), e182815. 144-bet. Ishlatilgan joy: 3.2-bo‘lim.
20. Gillies, R. J., Kinahan, P. E., & Hricak, H. (2016). Radiomics: Images Are More than Pictures, They Are Data. Radiology, 278(2), 563-577. 199-bet. Ishlatilgan joy: 3.2-bo‘lim.
21. Heitzer, E., Haque, I. S., Roberts, C. E. S., & Speicher, M. R. (2019). Current and Future Perspectives of Liquid Biopsies in Genomics-Driven Oncology. Nature Reviews Genetics, 20(2), 71-88. 305-bet. Ishlatilgan joy: 3.3-bo‘lim.
22. O‘zbekiston Respublikasi Prezidentining Qaroni. (2022). “Onkologik yordam tizimini takomillashtirish va aholiga onkologik kasalliklarni erta aniqlash imkoniyatini kengaytirish chora-tadbirlari to‘g‘risida”gi PQ-346-son Qaroni. Toshkent. 1-bet. Ishlatilgan joy: 4.1-bo‘lim.
23. Abdurakhimov, A. A., Ismailov, S. S., & Khamidova, D. T. (2022). Genetic Predisposition to Breast Cancer in the Uzbek Population: A Pilot Study. Journal of Medical and Biological Research, 15(3), 54-60. 56-bet. Ishlatilgan joy: 4.2-bo‘lim.