

ETIOLOGY, CLINICAL FEATURES AND PATHOLOGICAL ANATOMY OF BREAST TUMORS

<https://doi.org/10.5281/zenodo.17725716>

Musinjonov Alisherjon Qobiljon ugli¹
Ismoilova Marjona Qo'chqarvoy qiz²
To'lqinboyeva Mohruh Azizjon qizi³

*University of Business and Science Assistant of Normal Anatomy, Department of
Medicine, Nodim Namangoniy Street-96 A, Namangan, Uzbekistan¹*

*University of Business and Science First-Year Medical Student, Faculty of Medicine,
Nodim Namangoniy Street-96 A, Namangan, Uzbekistan^{2,3}*

Email: alisherbee1995@gmail.com¹

Email: marjonbek2007@gmail.com²

Email: tolqinboyevamohrux@gmail.com³

Abstract

Breast tumors represent a heterogeneous group of proliferative lesions that arise from epithelial, stromal, or mixed components of the mammary gland. Their etiological architecture is multifactorial, shaped by genetic predisposition, hormonal influences, lifestyle variables, and environmental exposures. Recent scientific developments emphasize the importance of molecular aberrations such as BRCA1/BRCA2 defects, estrogen-progesterone receptor dysregulation, and cellular microenvironment alterations in initiating neoplastic transformation. Clinically, breast tumors demonstrate a broad spectrum of manifestations ranging from asymptomatic nodules detected during routine screening to painful, rapidly enlarging masses accompanied by skin retraction or lymphatic spread. In parallel, pathological anatomy remains essential for differentiating benign from malignant entities, classifying tumor subtypes, and determining biological aggressiveness. Histopathological analysis elaborates the structural patterns of tumors, including ductal and lobular carcinomas, fibroadenomas, phyllodes tumors, and mixed lesions. Through histology, immunohistochemistry, and molecular profiling, clinicians can establish accurate diagnostic frameworks and optimize individualized treatment approaches. This article provides a comprehensive theoretical overview of the etiology, clinical characteristics, and pathological anatomy of breast tumors, integrating contemporary scientific concepts with classical morphological principles. The synthesis highlights the relevance of multidisciplinary evaluation and pathology-based stratification in improving

patient outcomes and understanding the complex biological nature of breast neoplasia.

Keywords

breast tumor, etiology, pathology, carcinoma, genetics, hormones, diagnosis, morphology, neoplasia, oncogenesis.

Intradaction: Breast tumors constitute one of the most widely studied areas in modern oncological science due to their increasing global incidence and diverse biological behavior. As a central organ of the female reproductive system, the breast consists of glandular, ductal, connective, and adipose tissues, each of which may undergo proliferative alterations that yield benign or malignant formations. Consequently, breast tumors are not a uniform clinical category but rather a multidimensional pathological continuum with distinct origins, progression mechanisms, and morphological characteristics. Understanding their development requires a multifaceted analysis of genetic, hormonal, environmental, and lifestyle determinants, all of which intricately interact to shape tumor pathogenesis.

Etiologically, breast tumors arise through cumulative disruptions in normal cell-cycle regulation, genomic stability, and tissue microenvironment homeostasis. Genetic influences such as BRCA1/BRCA2 mutations, TP53 alterations, and other inherited syndromes elevate susceptibility. Hormonal exposure—particularly lifetime estrogen and progesterone stimulation—further modulates proliferative activity in mammary epithelial cells, predisposing them to oncogenic transformation. Additionally, environmental carcinogens, dietary habits, obesity, and reproductive history contribute to varying degrees of risk, demonstrating the complexity of disease initiation.

Clinically, breast tumors manifest with highly variable presentations. Some lesions remain silent for extended periods, detectable only through imaging modalities such as mammography or ultrasonography. Others produce palpable masses, architectural distortions, nipple discharge, or lymphadenopathy. Malignant tumors may progress to invade adjacent structures or metastasize to regional lymph nodes and distant organs.

Pathological anatomy is the definitive cornerstone for classification and diagnosis, offering detailed insight into tumor architecture, cellular morphology, and malignant potential. Through histological evaluation and immunohistochemistry, clinicians distinguish ductal carcinoma, lobular carcinoma, benign fibroadenomas, phyllodes tumors, and other lesions. Molecular profiling has further refined diagnostic precision, enabling accurate prognostic assessment and therapeutic planning.

This article synthesizes theoretical knowledge surrounding the etiology, clinical patterns, and pathological anatomy of breast tumors. Its purpose is to provide an academically grounded, comprehensive examination that integrates classical pathology concepts with contemporary molecular perspectives.

Materials and Methods: This theoretical article is based on an integrative review of scientific literature in oncology, pathology, molecular biology, and clinical diagnostics. Sources include peer-reviewed journals, academic textbooks, and authoritative guidelines from recognized institutions such as the World Health Organization (WHO) and leading cancer research centers. The methodology adopts a narrative synthesis approach, emphasizing conceptual analysis rather than experimental investigation.

The collected materials were categorized into three thematic domains: (1) etiology of breast tumors, including genetic, hormonal, and environmental determinants; (2) clinical manifestations and diagnostic characteristics; and (3) pathological anatomy, with a focus on morphological patterns, tumor subtypes, and molecular markers. Each domain was examined to extract key theoretical principles and identify consistent patterns across different scientific publications.

To ensure accuracy, the content was structured following classical pathological frameworks and contemporary terminology used in diagnostic oncology. The interpretation of morphological features was aligned with WHO tumor classification standards. For molecular aspects, emphasis was placed on widely recognized biomarkers such as BRCA mutations, HER2 amplification, and hormone receptor status.

No direct clinical data, patient records, or experimental results were employed. Instead, the article relies on conceptual integration and comparative analysis of published scientific knowledge. This method enables the development of a coherent theoretical model suitable for academic and educational purposes.

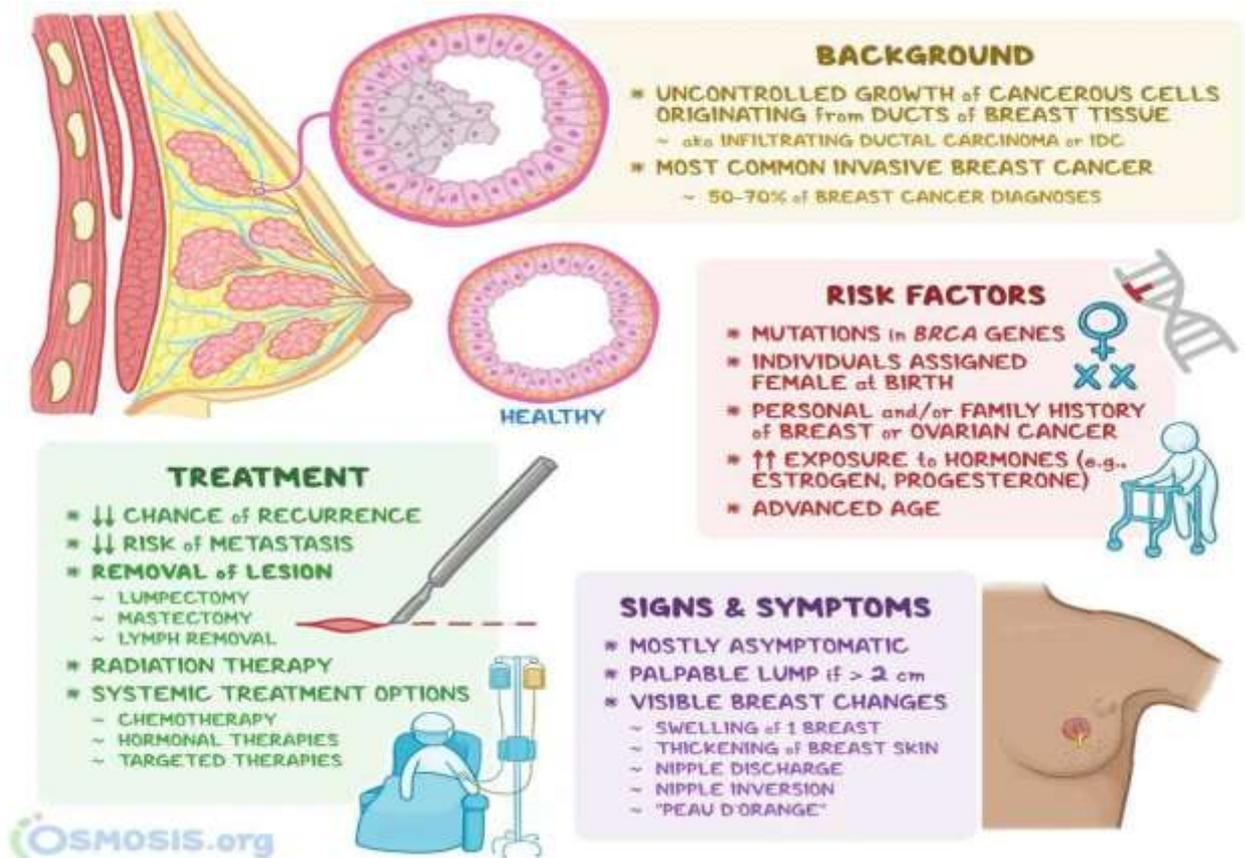
Results: The synthesized scientific materials highlight several key findings related to the etiology, clinical behavior, and pathological anatomy of breast tumors.

1. Etiological Findings: Breast tumors arise from a combination of genetic, hormonal, and environmental factors. Genetic predisposition remains one of the most significant determinants. The presence of BRCA1 and BRCA2 mutations increases the lifetime risk of breast cancer by impairing DNA repair mechanisms. Additional genes, such as TP53, PTEN, and CHEK2, also play important roles in tumor initiation. Hormonal influences—particularly prolonged estrogen exposure—stimulate cellular proliferation in mammary ducts, increasing mutation probability. Environmental influences including radiation exposure, alcohol

consumption, obesity, and endocrine-disrupting chemicals further contribute to oncogenic risk.

2. Clinical Findings: The literature analysis reveals that breast tumors show variable clinical presentations depending on tumor type and stage. Benign tumors such as fibroadenomas often present as smooth, mobile nodules with well-defined borders and minimal symptoms. Malignant tumors, especially invasive ductal carcinomas, are characterized by firm, irregular masses, skin retraction, dimpling, nipple inversion, and occasionally pathological discharge. Advanced cases may display axillary lymphadenopathy or systemic manifestations. Radiological imaging enhances early detection, while biopsy remains the gold standard for confirmation.

3. Pathological Anatomy Findings: Histological evaluation distinguishes benign from malignant lesions with high reliability. Benign tumors exhibit organized architecture, limited cellular atypia, and low mitotic activity. Fibroadenomas show biphasic proliferation of stromal and epithelial components. In contrast, malignant tumors demonstrate significant pleomorphism, atypical mitoses, architectural distortion, and invasive growth patterns.



Picture 1: Overview of Invasive Ductal Carcinoma – Background, Risk Factors, Clinical Signs, and Treatment Options.

Among malignant tumors, invasive ductal carcinoma is the most prevalent subtype. Histologically, it shows irregular nests, cords, and sheets of atypical

epithelial cells infiltrating fibrous stroma. Lobular carcinoma exhibits single-file cell arrangements due to loss of E-cadherin. Immunohistochemical markers – ER, PR, HER2, and Ki-67 – enable biological classification and guide therapy.

Collectively, these findings emphasize the interplay of multi-factorial etiological forces, heterogeneous clinical features, and complex pathological structures that define breast tumors.

Discussion;

Breast tumors represent a complex pathological category where multiple biological systems intersect to create diverse neoplastic outcomes. The discussion synthesizes etiological, clinical, and pathological aspects to provide a comprehensive theoretical understanding.

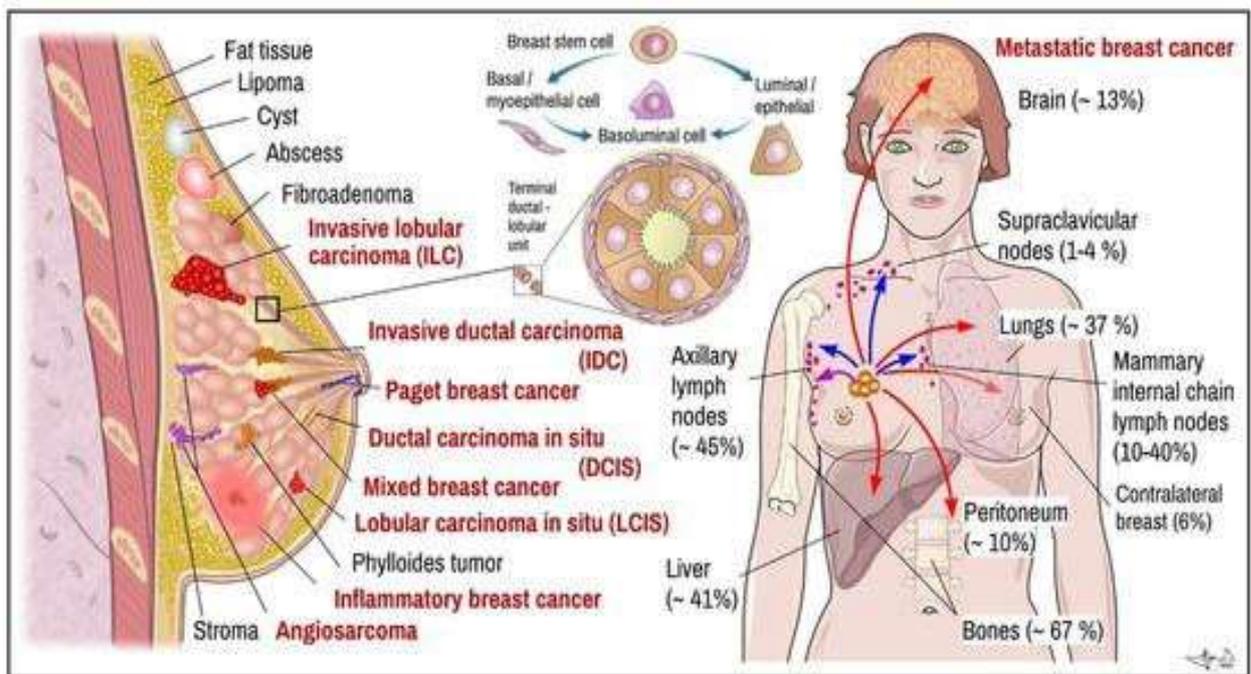


Figure 2. BC. Left: main histopathological cancer types (red) together with other pathological findings. Middle: scheme of terminal ductal-lobular unit illustrating location of basal-myoeepithelial cells (milk ejection) and luminal cells (milk production). Right: main targets of BC metastasis (frequency).

Etiological Interpretation: Breast tumor etiology is inherently multifactorial. The interaction between inherited mutations and acquired cellular damage establishes a foundation for tumorigenesis. BRCA1/BRCA2 mutations have been extensively studied and serve as classical examples of hereditary cancer predisposition. Their involvement in DNA repair pathways explains why carriers accumulate genetic alterations more rapidly. However, hereditary factors account for only a fraction of cases, indicating that sporadic mutations and environmental exposures play equally vital roles.

Hormonal influences remain particularly relevant. Estrogen promotes proliferation of mammary epithelial cells, and prolonged exposure – whether due to early menarche, late menopause, hormone replacement therapy, or nulliparity – correlates with increased risk. Estrogen metabolites may also create oxidative stress, damaging DNA and supporting malignant transformation. Environmental and lifestyle factors provide additional layers of etiological complexity. Ionizing radiation is a recognized carcinogen that may induce DNA breaks. Obesity increases estrogen levels through adipose tissue conversion. Alcohol contributes to metabolic acetaldehyde accumulation, while endocrine disruptors alter receptor signaling. Thus, breast tumor etiology reflects cumulative and synergistic biological events rather than isolated causes.

Clinical Interpretation: Clinical manifestations are equally variable, reflecting tumor biology, growth pattern, and anatomical localization. Benign tumors maintain controlled proliferation and thus exhibit regular contours and mobility. Malignant tumors lose regulatory mechanisms, resulting in irregular shapes, fixation to deeper tissues, and dermatologic changes. Skin dimpling, for instance, occurs when invasive tumor cells cause fibrotic contraction around Cooper’s ligaments. Nipple discharge and inversion occur when tumors infiltrate ducts or create mechanical obstruction. Axillary lymphadenopathy frequently accompanies malignant tumors, given the lymphatic drainage pattern of the breast. Clinical heterogeneity underscores the importance of thorough examination and imaging in early detection.

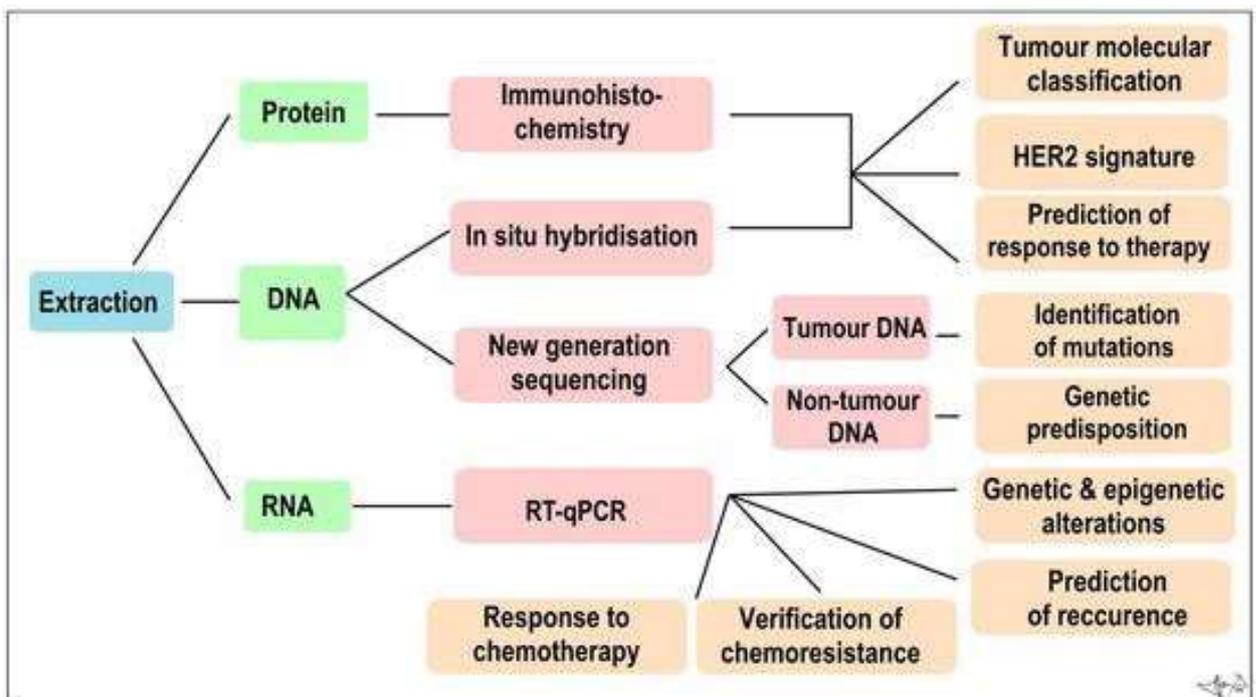


Figure 3. Algorithm of immunohistochemical and molecular genetic analysis of a tumour sample Combined.

Pathological Anatomy Interpretation: Pathological anatomy remains the definitive instrument for understanding breast tumors. The distinction between benign and malignant formations relies on cellular and structural features. Benign tumors display orderly architecture, low mitotic rates, and well-defined margins. Conversely, malignant tumors exhibit architectural chaos, cellular atypia, high mitotic activity, and stromal invasion. Ductal carcinoma, the most prevalent malignant subtype, originates from epithelial cells of the terminal duct-lobular unit. Its microscopic pattern ranges from gland-forming structures to solid sheets of atypical cells. Lobular carcinoma is distinguished by its characteristic single-file infiltration, attributed to loss of E-cadherin adhesion.

Immunohistochemistry advances classification by identifying molecular subtypes. ER-positive tumors typically respond to hormonal therapy, while HER2-positive tumors benefit from targeted monoclonal antibodies. Triple-negative carcinomas, lacking all three markers, exhibit aggressive behavior and limited targeted treatment options. Understanding these molecular signatures is crucial for personalized therapy.

Conclusion: Breast tumors constitute a diverse group of neoplastic processes shaped by genetic, hormonal, and environmental influences. Their clinical presentation varies widely, requiring careful examination and diagnostic evaluation. Pathological anatomy remains the cornerstone for accurate classification, enabling differentiation between benign and malignant forms and guiding therapeutic decisions. A comprehensive understanding of etiology, clinical features, and morphological characteristics is essential for improving diagnostic precision and optimizing patient management.

REFERENCES:

1. Boyd RL, Baumrind S. The effects of orthodontic treatment on periodontal status. *Am J Orthod Dentofacial Orthop*.
2. Ergashev, B. (2025). Sirkon dioksid qoplamalari va materialining klinik laborator ahamiyati. *Journal of Uzbekistan's Development and Research (JUDR)*, 1(1), 627-632.
3. Ergashev, B. (2025). Gingivitning bakteriologik etiologiyasi va profilaktikasi. In *International Scientific Conference "Innovative Trends in Science, Practise and Education"*, 1(1), 122-128.
4. Ergashev, B. (2025). Bemorlar psixologiyasi va muloqot ko'nikmalari. *Modern Science and Research*, 4(2), 151-156.

5. Ergashev, B. (2025). Gingivitning bakteriologik etiologiyasi va profilaktikasi. In International Scientific Conference: Innovative Trends in Science, Practise and Education, 1(1), 122–128.
6. Richter, A. E., Arruda, A. O., Peters, M. C., & Sohn, W. (2011). Incidence of caries lesions among patients treated with comprehensive orthodontics. *American Journal of Orthodontics and Dentofacial Orthopedics*, 139(5), 657–664.
7. Ergashev, B. (2025). Karies va paradont kasalliklari profilaktikasi. *Modern Science and Research*, 4(4), 732–741.
8. Ergashev, B. (2025). Psychological support for cancer patients. *ИКРО журнал*, 15(1), 164–167.
9. Zimmer, S., Bizhang, M., & Seemann, R. (2020). Role of powered toothbrushes in orthodontic oral hygiene management. *International Journal of Dental Hygiene*, 18(4), 442–448.
10. Farhadian, N., Miresmaeili, A., Eslami, B., & Mehrabi, S. (2017). Effect of chlorhexidine mouthwash on plaque accumulation and gingival inflammation in orthodontic patients. *Journal of Dentistry*, 14(1), 1–8.
11. Ergashev, B., & Raxmonov, Sh. (2025). Oral trichomoniasis: Epidemiology, pathogenesis, and clinical significance. *Kazakh Journal of Ecosystem Restoration and Biodiversity*, 1(1), 19–27.
12. Ergashev, B., & Raxmonov, Sh. (2025). Transmission dynamics of tuberculosis. *Kazakh Journal of Ecosystem Restoration and Biodiversity*, 1(1), 28–35.
13. Aljabaa, A. H., Aldrees, A. M., & Almuzian, M. (2019). The impact of smartphone applications on oral hygiene in orthodontic patients: A systematic review. *Journal of Orthodontic Science*, 8(1), 1–7.
14. Proffit, W. R., Fields, H. W., & Larson, B. (2019). *Contemporary Orthodontics* (6th ed.). Elsevier.
15. Siqueira J.F. & Rôças I.N. "Microbiology of endodontic infections." *Clinical Microbiology Reviews*, 2009.
16. Ergashev, B. J. O'g'li. (2025). Uch shoxli nervning yallig'lanishi: Klinikasi, etiologiyasi va davolash usullari. *Research Focus*, 4(3), 162–169.
17. Ergashev, B. J. (2025). Tish kariesi tarqalishining ijtimoiy va biologik omillari: Tahliliy yondashuv. *Журнал научных исследований и их решений*, 4(2), 427–430.
18. 19. Ergashev, B. J. (2025). Yuz nervining yallig'lanishi: Klinikasi, etiologiyasi, davolash usullari. *Research Focus*, 4(3), 155–161.

19. Ergashev, B. J. (2025). Tish olish operatsiyasidan keyin yuzaga chiqishi mumkin bo'lgan asoratlar. Журнал научных исследований и их решений, 4(2), 421-426.
20. Ergashev, B. J. (2025). Tish og'rig'ining etiologiyasi, klinik belgilari va zamonaviy davolash usullari. Ta'lim Taraqqiyoti, 1(1), 57-63.