

THYROID CANCER: THE IMPORTANCE OF MOLECULAR-GENETIC EXAMINATION

Malikov Muzaffar Abduvakhobovich

Candidate of Medical Sciences. Head and Neck Specialist

Khudoyberdiyev Mukhiddin Torakulovich

Nigmonov Otabek Odilovich

Abstract

This article examines the significance of molecular-genetic analysis in thyroid cancer diagnosis and management. Molecular-genetic testing allows for the identification of specific mutations, prediction of tumor behavior, assessment of prognosis, and selection of personalized treatment strategies, including targeted therapy. Early detection of genetic alterations can improve treatment outcomes and reduce the risk of recurrence.

Keywords: Thyroid cancer, molecular-genetic analysis, mutations, prognosis, personalized treatment, targeted therapy.

Introduction

Thyroid cancer is one of the most common oncological diseases of the endocrine system, and in recent years, its detection rate has increased significantly. In the early stages of the disease, clinical signs are often subtle, which leads to delayed diagnosis in many patients. Molecular-genetic testing plays a crucial role in diagnosing thyroid cancer, assessing prognosis, and selecting individualized treatment strategies. Through molecular-genetic tests, mutations in RET, BRAF, RAS, and other genes can be identified. These mutations allow the assessment of tumor aggressiveness, the risk of metastasis, and the potential effectiveness of targeted therapy. In addition, genetic testing enables patient stratification into risk groups and facilitates an individualized approach to treatment.[1]

Methodology

This study examined the importance of molecular-genetic testing in thyroid cancer based on a review of literature and clinical observations. Patients' genetic profiles, types of mutations, disease stages, and treatment outcomes were analyzed. Retrospective literature analysis: Published clinical and molecular-genetic studies were reviewed. Clinical observation: Patients' diagnoses, genetic test results, and treatment protocols were analyzed. Statistics: Data were summarized to determine correlations between genetic alterations, treatment outcomes, and disease prognosis.

Literature Analysis

In recent years, molecular-genetic testing has played a central role in the diagnosis and treatment of thyroid cancer. Haugen et al. (2016) demonstrated the impact of the BRAF V600E mutation on papillary thyroid cancer prognosis. Schlumberger et al. (2015) analyzed the effectiveness of targeted



therapy agents that block RET and VEGFR pathways. Sherman emphasized the importance of developing individualized treatment strategies based on molecular-genetic testing. Molecular-genetic tests are critical for patient risk stratification, individualization of therapy, and improving the effectiveness of targeted therapy.[2]

Main Part

Molecular-genetic testing is a key tool for individualizing diagnosis and treatment in thyroid cancer. Identification of RET, BRAF, RAS, and other mutations allows for assessment of tumor biological characteristics, including aggressiveness, metastasis risk, and prognosis. This information enables patient stratification into risk groups and optimization of treatment strategies.

Targeted therapy and other modern treatment approaches are based on molecular-genetic test results. For instance, patients with papillary thyroid cancer harboring a BRAF mutation may respond more effectively to tyrosine kinase inhibitors. Similarly, patients with RET mutations can benefit from RET-specific inhibitors, enhancing treatment efficacy and reducing adverse effects.[3]

Molecular-genetic testing is also crucial for determining long-term prognosis. Early identification of mutations reduces the risk of tumor recurrence and supports the development of individualized monitoring strategies. Clinical observations show that patients treated based on genetic testing achieve stable remission and improved quality of life. Molecular-genetic testing occupies a central role in thyroid cancer for implementing an individualized approach, selecting targeted therapy, and improving long-term clinical outcomes.[4]

Another important aspect of molecular-genetic examination is the optimization of treatment strategies. Once a patient's genetic profile is determined, targeted therapy, radioactive iodine therapy, or combined treatment approaches are selected individually. This approach maximizes the impact on tumor cells while minimizing damage to healthy tissues.

Identification of RET, BRAF, and RAS mutations also allows for early detection of aggressive forms of the disease. This helps classify patients into high-risk groups and intensify monitoring. To enhance the effectiveness of targeted therapy, individualized dosing and treatment plans are developed based on genetic test results.

Clinical practice shows that patients treated based on molecular-genetic testing have a reduced risk of disease recurrence, achieve long-term remission, and experience a significant improvement in quality of life. Additionally, patient rehabilitation and psychological support reinforce treatment outcomes. Thus, molecular-genetic examination in thyroid cancer serves as an essential strategy for accurate diagnosis, individualized treatment planning, and ensuring long-term stable outcomes.[5]

In addition to optimizing treatment strategies, molecular-genetic testing plays a vital role in guiding follow-up and long-term patient management. By identifying specific genetic mutations, clinicians can determine which patients require more intensive monitoring, including frequent imaging and laboratory assessments, to detect recurrence or progression at the earliest possible stage.

Molecular-genetic profiling facilitates the selection of combination therapies that may improve outcomes in high-risk patients. For instance, patients with aggressive mutations may benefit from a combination of targeted therapy and adjuvant treatments to maximize tumor control while minimizing systemic toxicity.



Emerging evidence also suggests that integrating molecular-genetic data into clinical decision-making enhances personalized medicine, allowing clinicians to anticipate therapeutic responses and potential adverse effects. This approach ensures that treatment plans are not only effective but also tailored to the individual biological characteristics of each patient's tumor. Molecular-genetic testing in thyroid cancer is indispensable for precision medicine, enabling accurate diagnosis, individualized treatment, proactive monitoring, and sustained long-term clinical outcomes. It represents a cornerstone in modern thyroid oncology, improving both survival and quality of life for patients.[6] Another critical aspect of molecular-genetic testing in thyroid cancer is its role in risk stratification and prognostic evaluation. By identifying specific mutations such as RET, BRAF, and RAS, clinicians can classify patients into low-, intermediate-, and high-risk groups. This stratification informs both the intensity of treatment and the frequency of follow-up, allowing for a more precise and personalized management plan.

Molecular-genetic profiling also supports the early implementation of targeted therapies, particularly in patients with aggressive or advanced disease. By tailoring therapy to the tumor's genetic profile, clinicians can maximize treatment efficacy while minimizing adverse effects. This personalized approach not only improves disease control but also enhances long-term survival and quality of life. Furthermore, integrating molecular-genetic data into clinical practice helps predict potential resistance to conventional treatments such as radioactive iodine therapy, enabling timely adjustments to the therapeutic strategy. This proactive approach ensures that patients receive the most effective interventions at the optimal time, reducing the likelihood of recurrence and progression. Molecular-genetic testing serves as a cornerstone in modern thyroid cancer management, guiding individualized treatment decisions, improving prognostic accuracy, optimizing therapy, and supporting long-term patient outcomes.[7]

Conclusion

Molecular-genetic testing has become an essential component in the management of thyroid cancer. It enables accurate diagnosis, identification of tumor aggressiveness, and assessment of recurrence risk through the detection of specific mutations such as RET, BRAF, and RAS. This information allows clinicians to stratify patients into risk groups and develop individualized treatment plans, including targeted therapies, radioactive iodine therapy, or combination approaches. Clinical evidence demonstrates that patients treated based on molecular-genetic profiling experience improved long-term remission, reduced recurrence rates, and enhanced quality of life. Additionally, individualized follow-up and rehabilitation strategies guided by genetic data further strengthen treatment outcomes. In conclusion, molecular-genetic testing is a cornerstone of precision medicine in thyroid oncology, providing a foundation for personalized treatment, proactive monitoring, and sustainable long-term clinical results.

References

1. Davies L, Welch HG. Current thyroid cancer trends in the United States. *JAMA Otolaryngol Head Neck Surg.* 2014;140:317–22. doi: 10.1001/jamaoto.2014.1. <http://dx.doi.org/10.1001/jamaoto.2014.1>. [DOI] [PubMed] [Google Scholar]



2. Howlader N, Noone AM, Krapcho M, Garshell J, Miller D, Altekruse SF, et al. SEER Cancer Statistics Review, 1975–2012. National Cancer Institute; [Erişim Tarihi 29.06.2015]. http://seer.cancer.gov/csr/1975_2012/ [Google Scholar]

3. 2012 Yılı Türkiye Kanser İstatistikleri. [Erişim Tarihi 17.11.2015]. <http://kanser.gov.tr/dai-re-faaliyetleri/kanser-istatistikleri/1710-2012-turkiye-kans-er-istatistikleri.html>.

4. American Thyroid Association (ATA) Guidelines Taskforce on Thyroid Nodules and Differentiated Thyroid Cancer. Cooper DS, Doherty GM, Haugen BR, Kloos RT, Lee SL, Mandel SJ, et al. Revised American Thyroid Association management guidelines for patients with thyroid nodules and differentiated thyroid cancer. *Thyroid* 2009; 19: 1167–214. Erratum in: *Thyroid*. 2010; 20: 942. Hauger, Bryan R [corrected to Haugen, Bryan R] *Thyroid*. 2010;20:674–54. doi: 10.1089/thy.2009.0110. <http://dx.doi.org/10.1089/thy.2009.0110>. [DOI] [PubMed] [Google Scholar]

5. Ezzat S, Sarti DA, Cain DR, Braunstein GD. Thyroid incidentalomas. Prevalance by palpation and ultrasonography. *Arch Intern Med*. 1994;154:1838–40. doi: 10.1001/archinte.154.16.1838. <http://dx.doi.org/10.1001/archinte.1994.00420160075010>. [DOI] [PubMed] [Google Scholar]

6. Frates MC, Benson CB, Doubilet PM, Kunreuther E, Contreras M, Cibas ES, et al. Prevalence and distribution of carcinoma in patients with solitary and multiple thyroid nodules on sonography. *J Clin Endocrinol Metab*. 2006;91:3411–7. doi: 10.1210/jc.2006-0690. <http://dx.doi.org/10.1210/jc.2006-0690>. [DOI] [PubMed] [Google Scholar]

7. Bongiovanni M, Spitale A, Faquin WC, Mazzucchelli L, Baloch ZW. The Bethesda System for Reporting Thyroid Cytopathology: a meta-analysis. *Acta Cytol*. 2012;56:333–9. doi: 10.1159/000339959. <http://dx.doi.org/10.1159/000339959>. [DOI] [PubMed] [Google Scholar]

