

# DIABETES AND ENDOCRINOLOGY

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## Molecular aspects of thyroid cancer

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Thyroid cancer constitutes a significant part of the malignant neoplasms of the endocrine system. Most tumors of this organ are of follicular cell origin and only 5-10% are parafollicular - C-cell (medullary) cancers. Currently, there are difficulties in the preoperative diagnosis of thyroid neoplasms. None of the currently employed methods have 100% specificity and sensitivity for the diagnosis of thyroid cancer. On the basis of a cytological study, it is not possible to distinguish, for example, follicular adenoma from follicular cancer, which is a significant problem when it comes to choosing the tactics of surgical treatment.

Thus, it is important to search for additional tools for preoperative differential diagnosis of thyroid neoplasms. A promising direction in this regard is the evaluation of the molecular genetics and expression profile of a thyroid tumor, the search for biomarkers that, in combination with a cytological study, would increase the accuracy of preoperative diagnosis of thyroid neoplasms.

The key and most studied events leading to the activation of mitogen-activated protein kinase (MAPK) signaling pathways that transduce myogenic signals and regulate cellular processes such as proliferation, differentiation and viability are mutations in the receptor (RET) and intracellular signaling molecules (BRAF and RAS). Mutations in the abovementioned genes are the most common tumor-initiating event in differentiated forms of thyroid cancer and are associated with specific clinical, histopathological and biological characteristics of tumors.

The most common somatic mutation in malignant neoplasms of the thyroid gland is V600E in the BRAF gene. The frequency of mutations in the BRAF gene in papillary thyroid

cancer is 40–45%, 1.3% in follicular thyroid cancers, 20-40% in poorly differentiated thyroid cancers and in anaplastic thyroid cancers the frequency is 30–40%. The frequency of mutations in codon 61 (Q61R, Q61K) in the NRAS gene in thyroid cancers is 6%, in codons 12, 13 and 61 in the KRAS gene - 3%.

Significant amount of evidence has been obtained by studying the expression profile of genes in thyroid tumors. An analysis of the literature data showed that the SDC4, PLCD3, PVRL4 genes are the most discriminating for the differentiation between benign and malignant neoplasms with a sensitivity and specificity of 100%, the SLC4A4 gene was the most effective marker in distinguishing between classic follicular cancer and the follicular variant of papillary cancer and papillary cancer.

Currently, on the basis of Moscow Clinical Scientific Center named after A. S. Loginov MHD, a number of somatic mutations and the expression profiles of thyroid neoplasms are being evaluated.

### Recent Publications

1. Bodunova N.A., Tsapkova L.A., Osipova (Savinkova) A.V. Possibilities of molecular genetic measurements in the differential diagnosis of thyroid neoplasms. // X Anniversary International Scientific and Practical Conference "Molecular Diagnostics 2021". - Moscow, p.154.

### Biography

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