

11. THE MOLECULAR PROFILE OF THYROID CARCINOMAS



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Introduction. Thyroid cancer is the most common endocrine malignancy, accounting for the seventh most common cancer in women. Thyroid nodules are potentially malignant. The determination of any mutation such as TERT, BRAF, PAX8/PPAR γ , RAS in a thyroid nodule provides a strong indication for malignancy.

Aim of the study. Studying the molecular profile for thyroid carcinomas

Methods and materials. An online database search of relevant published articles using the Cochrane Database of Systematic Reviews, PubMed, Embase and Google was performed via Google search.

Results. The specificity of the BRAF V600E PTC is 100%, but the sensitivity is 30%. BRAF mutations were found in 74% of PTC and 7.7% of FTC. RAS mutations are the primary changes in follicular adenomas up to 67%. The pAX/PPAR γ fusion rearrangement has an inactivating effect on the PPAR γ tumor suppressor gene and is found in 30–60% of FTC and 38% of the follicular variant of papillary thyroid cancer. TERTp mutations, more specifically C228T and C250T, account for 5–25% of PTC and 35% of FTC.

Conclusion. Molecular testing of thyroid nodules and thyroid cancer has improved the diagnostic accuracy of indeterminate thyroid nodules and provides useful information regarding tumor prognosis. Future development of predictive models that will combine genetic data with clinical and cytological findings will enable accurate preoperative risk assessment, precisely guiding individualized treatment options.

Keywords. Follicular thyroid cancer (FTC); molecular diagnosis; papillary thyroid cancer (PTC).