



# Genetic & Molecular Testing in Thyroid Cancer

**Genetic mutations may predispose you to tumor formation and cancer. Genetic testing can detect these mutations, confirm or rule out a diagnosis, and provide doctors with the information they need to design a targeted treatment plan.**

It is important to note that a final thyroid cancer diagnosis can only be given after the thyroid nodule has been surgically removed. However, ultrasound and fine-needle aspiration (FNA) biopsies can be incredibly useful in determining the best and least invasive treatment approach for you.

FNA biopsy is a minimally invasive sampling of a small number of cells from the thyroid nodule for microscopic examination. These biopsies can usually indicate whether a nodule is cancerous or not. In the event that results are still unclear, or 'indeterminate,' genetic testing can help clarify the risk of a nodule being malignant, or cancerous. Pre-established molecular markers can help predict the aggressiveness of the disease and the efficacy of various treatments.

## Types of Genetic Mutations

A variety of genetic mutations may be present in thyroid tumors. The most clinically relevant mutations include:

- **BRAF:** The most common mutation and often associated with aggressive thyroid cancers. BRAF mutations indicate a higher risk of recurrence and the likelihood of papillary thyroid carcinoma is nearly 100%.
- **RAS:** This mutation is typically associated with papillary or follicular thyroid carcinoma.



Please note that this information is intended for educational purposes. It does not replace consultation with your doctor, and it should not be interpreted as medical advice. We encourage you to speak to your health care provider if you have further questions or concerns regarding your medical care.

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- **RET/PTC:** This mutation is often associated with papillary and medullary thyroid carcinoma.
- **PAX8-PPAR:** This mutation is often associated with follicular thyroid carcinoma.

Once identified, doctors can use your specific genetic mutation to pursue more targeted treatment options. For example, specific drug combinations are far more effective against BRAF than other standard, non-targeted treatments.

## Genetic Testing Techniques

- **Polymerase Chain Reaction (PCR):** A method of amplifying a specific DNA sequence and identifying mutations by comparing it to a standard template code.
- **Sanger Dideoxy Sequencing:** This technique copies target DNA sequences to create DNA of different lengths, the ends of which are then tagged with fluorescent markers that can be used to detect single nucleotide variants.
- **Multiplex Ligation-Dependent Probe Amplification (MLPA):** A specific form of PCR that distinguishes between sequences differing by only a single nucleotide (point mutations).
- **Fluorescence *In Situ* Hybridization (FISH):** Tagging DNA with a fluorescent probe visualized using fluorescent microscopy.
- **Next-Generation Sequencing:** This method is used to rapidly sequence entire genomes at a relatively low cost.

## Who Should Get a Molecular Screening?

You should ask about molecular testing if two or more of your first degree relatives have had thyroid cancer, or if you have a neck mass that has undergone rapid growth.



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