

## **TERT Promotor Mutation as a Prognostic Indicator in Papillary Thyroid Carcinoma**

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**Introduction:** Papillary thyroid carcinoma (PTC) is considered a curable disease. However, not all PTC behaves the same, and tumor genetics offer an important tool for guiding clinical decision making. We present an interesting case of a patient thought to have intermediate risk PTC, who later presented with metastatic disease, associated with a high-risk mutation.

**Case Description:** A 69-year-old female with PTC presented for medical management after a total thyroidectomy. She received 105 mCi I-131 and a post therapy scan showed large uptake in the lower neck without distant metastasis. Whole body scan (WBS) one year later showed mild uptake in the lower neck. Tumor markers remained mildly elevated. She was lost to follow-up and presented later for evaluation of a new mandibular mass. A positron emission tomography (PET) scan showed bone metastasis in the left mandible as well as pulmonary metastases. Excisional biopsy confirmed metastatic PTC and genetic testing showed a telomerase reverse transcriptase (TERT) mutation. She had repeat I-131 therapy, which showed uptake in the mandibular mass. Her stimulated thyroglobulin (Tg) level was over 200 ng/mL. Her mandibular mass increased in size and she was referred to oncology. Tyrosine kinase inhibitor (TKI) chemotherapy was initiated. After two months, the mass regressed and Tg level improved to 39 ng/mL.

**Conclusion:** TERT promotor mutations in PTC were identified in 2013. They cause de novo formation of E-twenty-six (ETS) transcriptional factor binding sites, resulting in increased telomerase activity and causing malignant transformation. TERT mutation is associated with aggressive tumor behavior. Identification of these mutations can guide decision-making at the time of diagnosis. In our case, the complicated clinical course might have been anticipated if the mutation was initially known. This case highlights the importance of genetic alterations in PTC for prognosticating clinical trajectory and providing better patient care.