Germline mutations in cancer predisposition genes among patients with thyroid cancer.
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Abstract Text:

Background: Thyroid cancers are known component tumors of both well-described and emerging hereditary cancer syndromes. To assess the contribution of germline variants in thyroid cancer predisposition, we examined the prevalence of germline mutations among individuals with a history of thyroid cancer, compared to those with thyroid and breast cancer or breast cancer alone. Methods: Clinical histories and molecular results were reviewed for individuals with a history of thyroid and/or breast cancer, ascertained from a cohort of > 140,000 patients who underwent hereditary cancer multigene panel testing at a single commercial laboratory. Clinical history information was obtained from test requisition forms completed by ordering clinicians and from pedigrees/clinic notes, if provided. Results: Among 2,678 thyroid cancer patients, the majority were Caucasian (66.9%), female (92.3%), and/or had an additional cancer primary (71.9%), with nearly half reporting an additional breast cancer primary (49.1%). Among those with available pathology information, 4.1% had medullary thyroid cancer. The median (IQR) age at diagnosis was 38 (26,48) years, and while 94.1% had a family history of cancer, 78.8% had at least one affected 1st degree relative. Overall, 11.1% were identified as mutation carriers, defined as ≥1 pathogenic or likely pathogenic variant. Among those with thyroid cancer alone, 9.7% had a mutation, similar to those with breast cancer alone (9.7%) and those with breast and thyroid cancer only (10.5%). Genes most frequently mutated in the thyroid only group included CHEK2 (3.1%), MUTYH (monoallelic) (2.4%), APC (2.0%), ATM (1.6%), and PALB2 (1.2%). CHEK2 was the most frequently mutated gene observed in all groups, with a higher frequency seen among those with thyroid and breast cancer (5.5%) compared to breast cancer (2.5%) or thyroid cancer (3.1%) alone (p < 0.001). Conclusions: A high rate of germline mutations is observed among individuals with thyroid cancer presenting for clinical genetic testing, even in the absence of other primary cancer diagnoses. Thyroid cancer may be an under-recognized component tumor of hereditary cancer predisposition syndromes suggesting the need for further investigation.
Title:
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No

Is this abstract a clinical trial?
No

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No

Has this research been submitted for publication in a medical journal?
No

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