Thyroid Cancer, Nonmedullary, 5

Alternative Names
NMTC5

Record Category
Disease phenotype

WHO-ICD
Neoplasms > Malignant neoplasms

Incidence per 100,000 Live Births
Unknown

OMIM Number
616535

Mode of Inheritance
Autosomal dominant

Gene Map Locus
10q25.3

Description
Cancers of the thyroid gland can be divided into medullary thyroid cancers (MTC, derived from calcitonin-producing C cells) and nonmedullary thyroid cancers (NMTC, derived from follicular cells). Papillary cancers make up 85% of NMTCs, while the remaining are follicular or Hurthe cell cancers. NMTC5 is a nonmedullary cancer that runs in families. Symptoms of thyroid cancer include a painless lump in the neck, hoarseness of the voice, trouble swallowing, and a sore throat. NMTC is found to affect women more often than men. While it can occur at any age, the disease commonly presents itself in adults around the third or fourth decade of life.

For a definite diagnosis of thyroid cancer, a fine needle aspiration biopsy of the thyroid cells is usually carried out; thereby allowing pathologists to determine the exact type of cancer involved. Treatment involves a surgical thyroidectomy to remove the cancerous thyroid cells. This is followed by radioactive iodine treatment, a targeted chemotherapy approach that kills any remaining thyroid cells. Patients also require life-long thyroid hormone replacement. The drug levothyroxine is administered for this purpose. Prognosis of the disorder depends on the age and severity of the disease. However, compared to other types of thyroid cancer, NMTC is considered to be less aggressive and has a 97% cure rate.

Molecular Genetics
NMTC5 follows an autosomal dominant pattern of inheritance. It is believed to be caused by heterozygous mutations in the HABP2 gene. This gene encodes a serine protease that is speculated to act as a tumor suppressor. So far, only one variant; G534E (missense mutation), located within the serine-protease-trypsin domain of the HABP2 protein, has been linked to NMTC5. The mutation is predicted to cause a space constraint near the catalytic region, disrupting the active site and surface accessibility of its substrates.

Epidemiology in the Arab World

Saudi Arabia
Alzahrani et al. (2016) studied the association between HABP2 gene mutations and the risk of Nonmedullary Thyroid Cancer (NMTC) in Saudi Arabs. Eleven patients from four unrelated families affected by familial NMTC were recruited for the study. Genomic DNA isolated from the peripheral leucocytes of these patients did not find any mutations in the HABP2 gene. A total of 63 pediatric patients and 229 adult patients were then screened for the HABP2 mutation G534E using their paraffin embedded tumor tissues. The monoallelic mutation was found in one 33-year-old woman. She suffered from papillary thyroid cancer with extra-thyroidal invasion, lymph node metastasis, and multinodular goiter in the rest of the thyroid gland. In addition, 217 patients with sporadic NMTC were also screened using peripheral leucocytes and the G534E mutation was not found. Finally, 190 healthy controls were analyzed and the monoallelic mutation was found in one 27-year-old healthy male with no family history of thyroid illness. Hence, contrary to published reports, the authors found no strong relation between HABP2 and NMTC in a large Saudi cohort.

References

Alzahrani AS, Murugan AK, Qasem E, Al-Hindi H. HABP2 Gene Mutations Do Not Cause Familial or Sporadic Non-Medullary Thyroid Cancer in a Highly Inbred Middle Eastern Population. Thyroid. 2016; 26(5):667-71. PMID: 26906432.

Related CTGA Records

Hyaluronan-Binding Protein 2

External Links

http://www.cancer.org/cancer/thyroidcancer/index
http://www.nhs.uk/conditions/Cancer-of-the-thyroid/Pages/Introduction.aspx
https://www.cancer.gov/types/thyroid/patient/thyroid-treatment-pdq

Contributors

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