Medullary Thyroid Cancer and the Impact of Genetic Testing

Erika L. Keatts, MSN, RN, and Joanne Itano, RN, PhD, OCN®

Medullary thyroid cancer (MTC) consists of a rare, undifferentiated tumor and often is described as having a chronic and indolent disease process. Approximately 5%–10% of all thyroid malignancies are MTC, and about 25% of patients diagnosed with the disease have a genetic form that was inherited through a mutation of the RET proto-oncogene. The mutation is expressed by an autosomal dominant allele and, if inherited, has almost a 100% chance of developing into a malignancy. Detection of the germline mutation identifies individuals at risk and enables prophylactic treatment for the prevention of MTC. As a result, patients and family members commonly undergo genetic testing during the diagnostic phase and experience certain psychosocial stressors. The purpose of this article is to provide an overview of MTC and its symptoms, treatment, prognosis, and genetics. The psychosocial effects of genetic testing on the quality of life of patients with MTC also will be described. By learning more about the pathophysiology and psychosocial stressors, nurses can facilitate proper counseling and increase the likelihood of positive outcomes for their patients.

At a Glance
✦ Medullary thyroid cancer (MTC) is a cancer of the parafollicular cells of the thyroid gland. Although 75% of MTC cases are sporadic and develop without genetic involvement, 25% are genetically inherited.
✦ A child born to a parent with familial MTC has a 50% chance of inheriting the germline mutation of the RET proto-oncogene, which, if inherited, has almost a 100% chance of developing into the malignancy.
✦ Addressing issues related to genetic testing can enable healthcare providers to assist patients in adjusting to psychosocial stressors.

Cancer Society (2006) estimated that 30,180 people will be diagnosed with thyroid cancer in 2006. Six percent of all women and 1.5% of all men will develop a single palpable thyroid nodule.

Thyroid Cancer

Thyroid cancer represents only a small percentage of malignancies diagnosed in the United States. In fact, the American...