

CJON WRITING MENTORSHIP PROGRAM PAPER

Medullary Thyroid Cancer and the Impact of Genetic Testing

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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.

Medullary thyroid cancer (MTC) accounts for 5%–10% of all thyroid malignancies (Van Nostrand, Bloom, & Wartofsky, 2004). A rare, undifferentiated tumor, MTC often is described as having a chronic and indolent disease process because it progresses slowly, over years or decades, with or without symptoms. Like other chronic conditions, MTC has the potential to significantly impact diagnosed individuals and their loved ones. The effects may be physical as well as emotional, social, and sometimes even financial. Collectively, the effects of MTC can perpetuate a change in well-being, or quality of life (QOL), in patients who are diagnosed with the disease.

A unique characteristic of MTC is its genetic transmission. Approximately 75% of MTC cases are sporadic and develop without genetic involvement (Van Nostrand et al., 2004). The remaining 25% of cases are considered an inherited form of MTC. The familial form of MTC is expressed by an autosomal dominant allele and appears when a mutation occurs in the RET proto-oncogene (Samaan, Ordonez, & Hickey, 1993). The mutations cause direct abnormal cell growth and tumor formation.

Unfortunately, literature about MTC is limited. The purpose of this article is to provide an overview of MTC and its symptoms, treatment, prognosis, and genetics, including a description of psychosocial effects related to genetic testing on the QOL of patients with MTC.

Thyroid Cancer

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

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

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