Medullary Thyroid Cancer and the Impact of Genetic Testing

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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 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.