Multiple endocrine neoplasia 2 (MEN2) is a hereditary syndrome associated with medullary thyroid carcinoma, pheochromocytoma, and hyperparathyroidism. Unfortunately, a diagnosis of MEN2 often is delayed until after the patient has developed an advanced MEN2-related tumor. Nurses should be familiar with hallmark signs of this syndrome to facilitate an early diagnosis and appropriately refer families for genetic assessment and, ultimately, develop a long-term plan for early detection and intervention for all family members at risk for developing MEN2.

T.M. is a 63-year-old man who presented to his primary care physician with a six-month history of chronic fatigue. Initial testing done by his physician revealed hypercalcemia. T.M. was referred to an endocrinologist, and additional testing demonstrated elevated calcitonin levels, which often are indicative of thyroid dysfunction, either C-cell hyperplasia or medullary thyroid carcinoma. He also had an elevated parathyroid hormone level. Ultrasound evaluation revealed a 1.8 cm x 1.5 cm nodule in the left lobe of the thyroid. A nuclear thyroid scan was ordered and T.M. was referred to the oncology clinic for additional evaluation.

Nursing Assessment and Physical Examination

On assessment, the nurse reviewed T.M.’s past medical and family history. T.M. had a history of depression and hyperlipidemia. A palpable thyroid nodule was noted on physical examination. He complained of recurring episodes of diarrhea and neck pain. Past surgeries included cholecystectomy and a hernia repair. The family history is notable for a paternal uncle who was diagnosed with thyroid cancer at age 45 (see Figure 1). T.M.’s father died at age 39 in a motor vehicle accident. T.M. is married and has a 38-year-old daughter and three sons (ages 40, 36, and 34 years). His daughter has thyroid problems of an unknown etiology. T.M. has 10 grandchildren. He works as a truck driver and is frustrated that he has had to take numerous sick days because of his chronic fatigue.

In T.M.’s case, the combination of hyperparathyroidism, elevated calcitonin levels, and the family history of thyroid cancer was highly suggestive of multiple endocrine neoplasia 2 (MEN2). In light of these findings, T.M. was referred for genetic evaluation and testing that identified a mutation in the RET gene consistent with level 2 (intermediate risk) MEN2. Based on the test results, T.M. underwent a total thyroidectomy with cervical lymph node dissection. Parathyroid exploration revealed a large mass which was excised. Pathology results confirmed bilateral medullary thyroid carcinoma with cervical lymph node metastases and a parathyroid adenoma. T.M.’s daughter and two of his sons (the 34- and 36-year-olds), as well as four grandchildren younger than age 16, also tested positive for the same RET mutation. All family members who tested positive for the RET mutation subsequently underwent prophylactic thyroidectomies. They continue to undergo surveillance for residual thyroid carcinoma, pheochromocytomas (a rare tumor of the adrenal gland), and hyperparathyroidism. (See Figure 2 for descriptions of common problems associated with MEN2.)

Etiology of the Problem

Medullary thyroid cancer (MTC) comprises 3%–10% of all thyroid cancers (Lindor, McMaster, Lindor, & Greene, 2008). Patients with MTC often present with a palpable thyroid nodule. Symptoms that may be associated with MTC include hoarseness, difficulty swallowing, dyspnea, and pain in the throat or neck. Diarrhea, resulting from elevated calcitonin levels, also can be a presenting symptom (Moline & Eng, 2010).

About 25%–30% of MTC malignancies are caused by MEN2 (Moline & Eng, 2010). MEN2 is an autosomal-dominant