Case Reports

Neurofibroma Adjacent to the Thyroid Gland and a Thyroid Papillary Carcinoma in a Patient with Neurofibromatosis Type 1: Report of a Case

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Abstract

Neurofibromatosis (NF) type 1, also known as von Recklinghausen's disease, is an autosomal-dominant inherited disorder. Some tumors may develop in these patients, including optic pathway gliomas, astrocytomas, brainstem gliomas, chronic myeloid leukemia, and rhabdomyosarcoma. Patients with neurofibromatosis type 1 show an increased risk of endocrine tumors, especially pheochromocytomas, whereas thyroid carcinoma is very rare. It is also rare for a neurofibroma to arise in the tissue neighboring the thyroid gland, and mimicking a nonfunctional thyroid nodule. This report presents a case of a neurofibroma adherent to the thyroid gland with thyroid papillary carcinoma in a 26-year-old woman with NF type 1.

Key words Neurofibromatosis type 1 · Thyroid papillary carcinoma · Neurofibroma

Introduction

Neurofibromatosis (NF) type 1 is an autosomal-dominant inherited neurocutaneous disorder. Café-au-lait patches, neurofibromas, skin-fold freckling, Lisch nodules, optic pathway glioma, and bony dysplasia are major diagnostic features of NF type 1. The Nf1 gene is located on chromosome 17q11.2 and the product of this gene, neurofibromin, acts as a tumor suppressor protein. In these patients some tumors may develop, including optic pathway gliomas, astrocytomas, brainstem gliomas, chronic myeloid leukemia, rhabdomyosarcoma, and endocrine tumors, especially pheochromocytoma. Thyroid carcinomas including papillary carcinoma are very rare in NF type 1.

Another rare condition is a neurofibroma in thyroid tissue mimicking a nonfunctional thyroid nodule. This report presents the case of a neurofibroma adherent to the thyroid gland with thyroid papillary carcinoma in a 26-year-old woman with NF type 1.

Case Report

A 26-year-old woman with NF type 1 was admitted because of a large mass located on her neck for the previous 2 years. Her family history was unremarkable. A physical examination revealed multiple café-au-lait patches and neurofibromas all over the body surface, skin-fold freckling, an enlarged thyroid gland, and a 7 × 6-cm nodule placed on the right thyroid lobe (Fig. 1). A complete blood count, serum biochemistry, and urinalysis were normal. The patient was euthyroid, with serum levels of free T4, 1.51 ng/dl (normal: 0.85–1.78); thyroid-stimulating hormone, 2.53 μIU/ml (normal: 0.4–4); and thyroglobulin, >300 ng/ml (normal: 0–55). Autoimmune antibodies such as antithyroglobulin and antimicrosomal antibodies were within the normal limits. Thyroid ultrasonography (US) revealed a 30 × 15-mm hypoechoic and heterogeneous nodule in the isthmus and heterogeneous parenchyma of thyroid gland. In addition, a well-defined, hypoechoic solid mass, 75 × 64 × 32 mm in size, adherent to the right lobe of the thyroid gland, was detected on US examination. Abdominal US and magnetic resonance imaging (MRI) of brain for NF type 1 were normal. The patient underwent surgery for multinodular goiter and a solid mass adherent to the right lobe of the thyroid gland; a total thyroidectomy was performed and the adherent mass was totally removed.

The tumors in the thyroid gland were unencapsulated, solid, and demonstrated a gray-yellow cut surface. Microscopically, they were dense and contained pink-staining colloid, clear, overlapping nuclei, and nuclear...
The tumor was diagnosed as a follicular variant of papillary thyroid carcinoma. On microscopic examination, the solid mass adherent to the right lobe of thyroid gland consisted of spindle-shaped cells with wavy and tapered nuclei; the cells showed positive staining for S-100 and a neurofibroma was diagnosed (Fig. 3). Radioactive iodine treatment was performed for adjuvant therapy of papillary carcinoma. At present, the patient is alive without any evidence of recurrence or metastasis 4 months postoperatively.

**Discussion**

Neurofibromatosis type 1 is a type of phakomatosis inherited in an autosomal-dominant fashion. It is characterized by: (1) six or more café-au-lait spots over 5 mm in their greatest diameter during the prepubertal age and over 15 mm in their greatest diameter during the postpubertal age, (2) axillary or inguinal freckling, (3) Lisch nodules, (4) two or more neurofibromas or one plexiform neurofibroma, (5) osseous lesions such as