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Visual Vignette

A Patient With Cowden Syndrome Presenting With a Multi-Nodular Goiter

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Case Presentation

A 33-year-old man presented for evaluation of a large multinodular goiter. One of the thyroid nodules was biopsied previouslyand showed atypia of undetermined significance. Thyroidectomy was recommended but the patient requested a second opinion. His medical history was significant for acral junctional nevi, melanoma, and skin tags. The physical exam was significant for macrocephaly (66 cm); several oral papillomas on the buccal mucosa (Fig. 1), inner lips, and tongue, which gave rise to a cobblestone appearance; palmoplantar keratotic pits (Fig. 2); and multiple lipomas and cutaneous facial papules. The thyroid was markedly enlarged with multiple nodules. Serum thyroid stimulating hormone level was normal. We biopsied the previously biopsied nodule, which again showed atypia of undetermined significance. The genomic sequencing classifier on the nodule was negative. The patient declined thyroidectomy and chose to monitor the nodules with yearly neck ultrasound. Given his physical findings, he underwent genetic testing, which identified the pathogenic mutation of *PTEN* gene c.388C>T (p.Arg130*). None of his first-degree relatives share a similar phenotype. This suggests that he is the first in the family with such *PTEN* mutation.

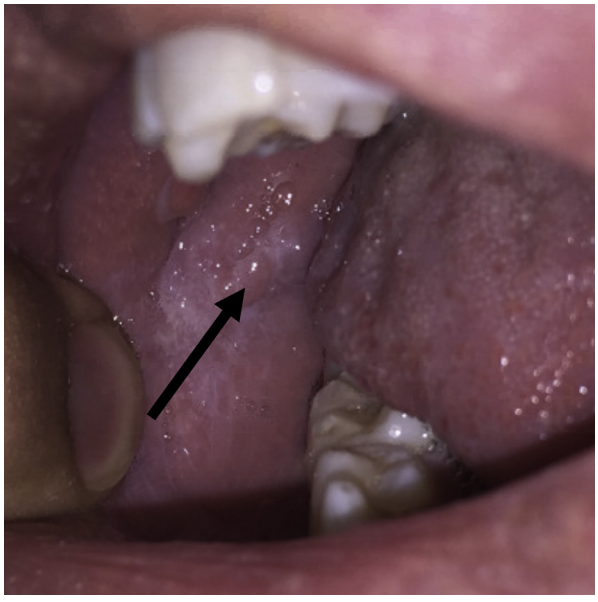

Fig. 1.

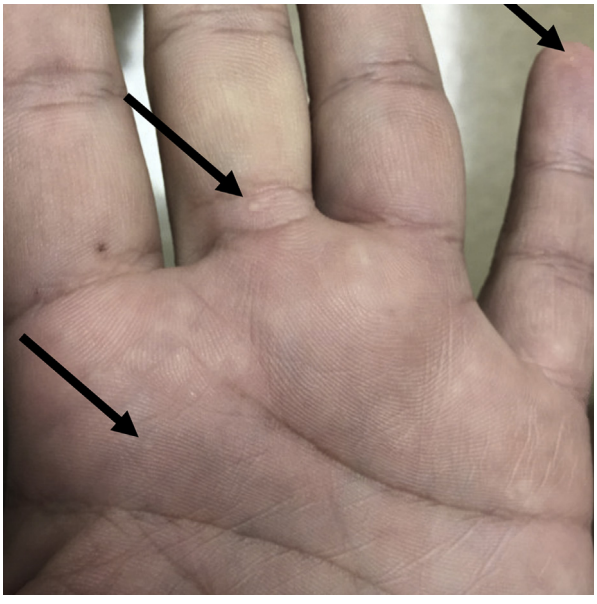

Fig. 2.

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What is the diagnosis?

Answer

Cowden syndrome (CS). CS is one of several autosomal dominant-inherited genetic syndromes resulting from germline mutations of the tumor suppressor *PTEN*. These mutations produce abnormal PTEN enzyme that fails to restrain cell proliferation or induce apoptosis. CS is characterized by multiple hamartomas and increased risk of thyroid, breast, and endometrial cancer. Pathognomonic mucocutaneous lesions of CS include oral mucosal papillomatosis, palmoplantar keratosis, and trichilemmoma. Macrocephaly and multiple lipomas are common features in CS. Patients with CS are at risk of early-onset thyroid neoplasm, with a 35%-38% life-time risk of thyroid cancer and a 9-fold risk of pediatric-onset thyroid cancer.[1] The median age of the patient at thyroid cancer diagnosis was 35.[2] Follicular thyroid cancer is the most common thyroid cancer in CS. The management of CS centers on the early detection of cancer. Current recommendations include yearly thyroid ultrasound and dermatologic examinations. Screening for thyroid cancer should be performed at the time of the diagnosis, regardless of age. Biennial colonoscopy should begin at age 40. For female patients, yearly mammogram and transvaginal ultrasound are recommended.[3] This case highlights the need to recognize the aforementioned physical findings of CS in patients who present with thyroid nodules. Recognition of CS provides the opportunity for earlier surveillance and possible prophylactic intervention for its associated malignancy.

Disclosure

The authors have no multiplicity of interest to disclose.

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