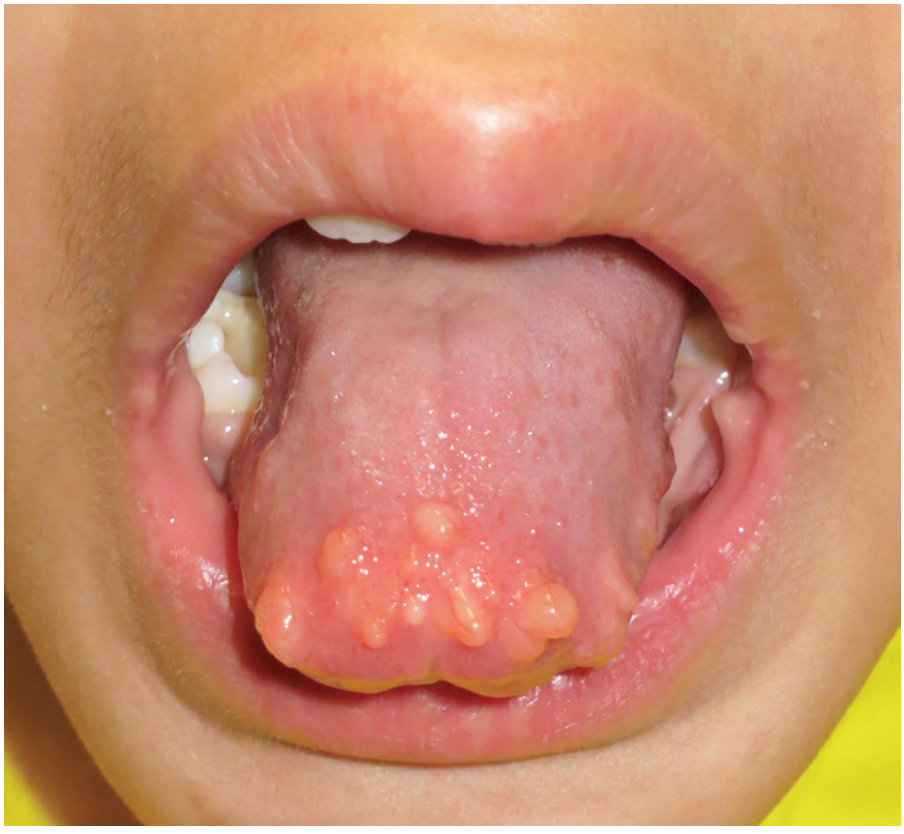
IMAGES IN CLINICAL MEDICINE

Chana A. Sacks, M.D., *Editor*

Mucosal Neuromas



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A 7-year-old boy presented to the pediatric otolaryngology clinic with a 3-year history of multiple firm, painless, slow-growing nodules on his tongue. He had a history of mild developmental delay, and a physical examination showed a high arched palate and marfanoid habitus. Excisional biopsies of the lesions on the tongue revealed mucosal neuromas. Multiple endocrine neoplasia type 2B was suspected. Ultrasonography of the neck revealed hypoechoic thyroid nodules without cervical lymphadenopathy. The calcitonin level was 407 ng per liter (normal value, <6); serum levels of metanephrines were within the normal range. There was no family history of multiple endocrine neoplasia. Genetic testing confirmed a pathogenic mutation in the gene encoding ret proto-oncogene (*RET*). Multiple endocrine neoplasia type 2B is an autosomal-dominant syndrome characterized by mucosal neuromas, pheochromocytoma, and medullary thyroid carcinoma. Early diagnosis of medullary thyroid cancer is critical to improving long-term outcomes; therefore, the recognition of nonendocrine manifestations is important in this syndrome. The child underwent a total thyroidectomy and bilateral neck dissection, which revealed three separate foci of medullary microcarcinoma without extrathyroidal spread. At a 9-month follow-up visit, the child had recovered well from surgery and was growing normally. After the visit, he continued to undergo routine clinical surveillance.

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