Rare Mucosal Neuroma on the Basis of Language: Case Report

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Introduction

It is believed that the mucosal neuroma is considered pathognomonic of multiple endocrine neoplasia type 2B (MEN 2B) and is observed in almost 100% of the patients. They are usually present at birth or in childhood and occur mainly in the lips, tongue, oral cavity, eyelids and gastrointestinal tract. It is present mainly between the first and second decade of life. Multiple endocrine neoplasia type 2B (MEN 2B) is a rare autosomal dominant syndrome composed of medullary thyroid carcinomas, pheochromocytoma and various mucosal neuromas [1]. The point mutation (ATG for ACG) of the RET proto-oncogene, which encodes a tyrosine kinase receptor expressed in neural crest-derived tissues, has been observed in approximately 90% of MEN 2B patients. However, multiple solitary neuroma without MEN 2B is an extremely rare case, only four cases occurring in the conjunctiva, bronchi, tongue and rectosigmoid colon are reported.

Clinically, the mucosal neuroma presents as pedunculated nodules or papules on the anterior, lateral and dorsal surface of the tongue. Other features that may alert clinicians to a diagnosis of MEN 2B include finding the thickened corneal nerve on top of an ophthalmologic assessment and the presence of skeletal manifestations such as scoliosis, kyphosis, lordosis, ligament laxity, slippage of femoral epiphyses. The complete syndrome with mucosal neuroma, pheochromocytoma and medullary thyroid carcinoma occurs in only 50% of cases and when present it is too late for a prophylactic approach [2]. The identification of early manifestations such as intestinal ganglioneuromatosis and oral mucosal neuroma should alert the physician to initiate an investigation for multiple endocrine neoplasia type 2B.

Early diagnosis is important because they usually precede thyroid and adrenal neoplasms. But it is often done late, especially when there is no family history of MEN 2B. In newborns, the MEN 2B phenotype characteristic is often absent and the only common manifestation is constipation due to intestinal ganglioneuromatosis. It is suggested that annual laboratory tests be performed from 3 years to 35 years of age in all patients with mucocutaneous characteristics suggestive of MEN 2B and in first-degree relatives of affected persons. The diagnosis of MEN 2B is also necessary to provide appropriate investigation of associated diseases and to enable counseling and screening of relatives for a mutation. Even patients with MEN 2B, who often have typical physical characteristics, may not be properly recognized and followed as a sporadic case. Based on this, all suspected cases of multiple endocrine neoplasia should undergo a molecular genetic test.

Case Report

A 56-year-old female patient reported the appearance of a nodule on the base of the left tongue with an evolution of approximately 8 months, progressive growth and sporadic local pain. Negative weight loss in this period he stopped smoking 3 months ago. At physical examination, the oroscopy had a firm and hardened nodule, with no ulcerated appearance of 2.4 x 2.2 cm in the base of the left tongue. First it was suspected of ectopic thyroid; however tests showed thyroid in normal topography head and neck without nodules or palpable masses [3]. Magnetic resonance imaging showed a nodular lesion approximately 3 cm in diameter on the base of the tongue and left lateral border. Cervical neck and US scintigraphy were normal. A biopsy of the nodule that presented the result of mucosal neuroma was performed. The patient was referred to the endocrinology department and maintained at the otorhinolaryngology outpatient clinic of the Santa Casa de Misericórdia hospital in Curitiba.

Discussion

The mucosal neuroma is considered a true neuroma, which is found in multiple endocrine neoplasia type 2B (NEM 2B). It can be confused with a post-traumatic neuroma, so it is important during the anamnesis to have the patient’s previous
morbid history, in order to be able to discard this diagnostic possibility. In the patient in question, this relationship was not found with multiple endocrine neoplasia type 2B, differently from most cases found in the literature. The mucosal neuroma is more epidemiologically found in places such as the lips, tongue, oral cavity, eyelids and gastrointestinal tract [4]. As is the case of the patient in question, in which the mucosal neuroma is located at the base of the tongue. Another epidemiological data found in the literature is the prevalence of mucosal neuroma in males and young people, different from the patient reported in the case, who is a middle-aged woman.

It is important that the patient is screened for NEM 2B, through a genetic screening, considering the great possibility of association of this syndrome with the pathology in question. Genetic screening should involve the search for mutations in the proto-oncogene RET, autosomal dominant inheritance, since most of the affected patients present the mutation M918T in exon 16 of RET. The diagnosis of the mucosal neuroma should be performed as early as possible, since it may precede the appearance of neoplasias, such as the thyroid or adrenal glands. Often it is necessary to perform prophylactic surgery, such as thyroidectomy to prevent these neoplasms. In the patient in question, imaging tests such as magnetic resonance imaging and thyroid ultrasonography were performed for evaluation.

**Conclusion**

There is a high prevalence of patients with mucosal neuroma that are associated with multiple endocrine neoplasia type 2B. Patients with this condition should be investigated and screened to rule out such syndrome. The earlier the diagnosis is made, the better the patient’s prognosis. Prophylactic procedures for the prevention of associated neoplasms are often necessary.

**References**

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