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Case Report

Multiple Endocrine Neoplasia Type 2B: Early Diagnosis by Multiple Mucosal Neuroma

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Abstract

Multiple Endocrine Neoplasia Type 2B (MEN 2B) is an uncommon autosomal dominant disorder characterized by the presence of medullary thyroid carcinoma, pheochromocytoma, and mucosal neuromas that can develop in various locations such as the tongue, lip, intestinal tract, and palate. They typically emerge in early childhood. Early identification and appropriate management are crucial in minimizing the progression of this discorder. This article reports a -year-old male patient with mucosal neuromas and previous history of medullary thyroid carcinoma.

Keywords: Medullary thyroid carcinoma; MEN 2B; Multiple mucosal neuroma

Introduction

Multiple Endocrine Neoplasia Type 2 (MEN 2) is a rare hereditary disorder that can be categorized into three distinct subtypes: MEN 2A, MEN 2B, and familial medullary thyroid cancer (FMTC) [1,2]. Among these, MEN 2B is characterized by the presence of -medullary thyroid cancer (MTC), pheochromocytoma, mucosal neuroma, ganglioneuromatosis, and a Marfanoid appearance [3].

Mucosal neuroma is a distinctive phenotype of MEN 2B, typically emerging at birth or around one to two years of age. Consequently, the early detection of mucosal neuroma is vital for a favorable prognosis and serves as a dermatologic indicator for diagnosis. In the local dermatologic literature, we encountered a case of MEN 2B initially diagnosed as MTC in the internal medicine department, with subsequent consultation with the dermatology department due to multiple papules on the lips and tongue [4].

In this report, we present an intriguing case of a 23-year-old male who exhibited multiple verrucous papules and nodules on his lips, tongue, and gingiva since childhood and received consequently an early diagnosis of MEN 2B.

Case Presentation

A 26-year-old male presented with the chief complaint of asymptomatic increased lip volume that evolved since childhood. Extraoral examination revealed multiple nodules on the conjunctival surfaces of the upper and lower eyelids. The lips were diffusely enlarged and appeared everted and bumpy on palpation. On intraoral examination, multiple nodules were seen on the tip and lateral borders of the tongue that appeared enlarged with dental misalignment. Nodular masses were also seen on the buccal mucosa and labial mucosa. The nodules were painless, oval-to-round shape, measuring 8mm × 5 mm in dimension with sessile base and smooth surface **(Figure 1)**. Anterior maxillary and mandibular teeth appeared to be protruded from side profile view **(Figure 2)**. His past medical history showed severe constipation since childhood but the family history showed no specific endocrine disease. He also gave a history of medullary thyroid carcinoma 02 years back.

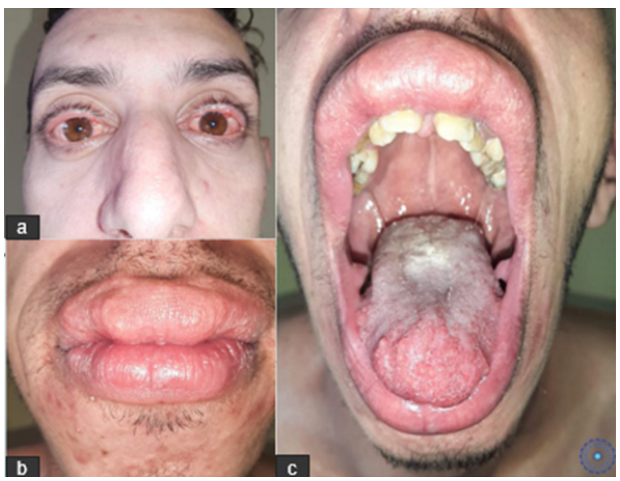

Figure 1: (a) Neuromas in the eyelids (b) Enlarged bilateral lips (c) Neuromas in the buccal mucosa with dental misalignment.

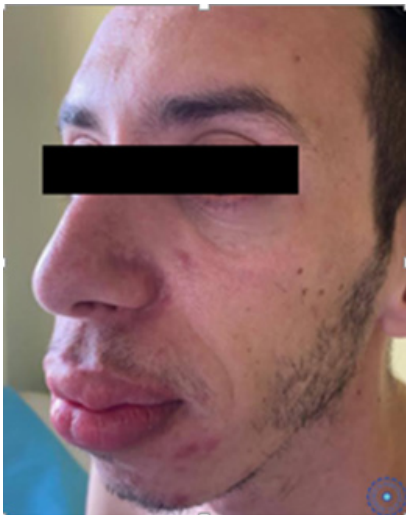

Figure 2: Side profile of the patient (Anterior protrusion of maxillary and mandibular teeth).

Incisional biopsy was done from nodule on labial mucosa. Histopathological examination revealed bundles of disorganized and tortuous nerve fibers surrounded by a thickened perineurium which was suggestive of mucosal neuromas. Based on the clinical, histological features and history, a clinical diagnosis of MEN2B was made. Since none of the family members were affected, this case was considered to be due to de novo mutation. The patient was further referred to the Department of Endocrinology for further management.

Discussion

MEN 2B accounts for just 5% of all MEN 2 cases; nevertheless, it exhibits the most aggressive clinical course [5]. While sharing similarities with MEN 2A, MEN 2B is distinguished by the presence of mucosal neuromas, ganglioneuromatosis in the intestinal tract, and a Marfanoid habitus, features not observed in MEN 2A.

Mucocutaneous neuromas appearing on the tongue and subconjunctiva during early childhood could represent some of the earliest manifestations and serve as clinical indicators for diagnosing this condition. The characteristic clinical features of this cutaneous neuromas include lip enlargement, the presence of multiple mucosal neuromas on the eyelids, lips, tongue, and buccal mucosa. In our case, additional intraoral features were observed, including a high-arched palate, gingival hyperplasia, prognathic mandible, and spacing of anterior teeth. Pheochromocytoma is present in about 50% of MEN2B patients [6]; however, it was not yet found in the present case. Most patients exhibit intestinal ganglioneuromatosis, with MTC being the next common component that may have an early onset.

Since the majority of MEN2B cases result from de novo mutations, diagnosing this condition can be challenging for a general practitioner. Therefore, they should specifically search for key diagnostic indicators, including mucosal neuromas on the tongue, lips, buccal mucosa, and inner eyelids [7].

The diagnosis in the current case was established based on the observation of a marfanoid habitus, the presence of multiple submucosal neuromas, a documented history of medullary thyroid carcinoma, and histopathological confirmation of mucosal neuroma.

Conclusion

Early diagnosis is essential, as many patients develop thyroid malignancy. MEN2B is of particular interest to dermatologists, as cutaneous and mucosal neuromas are among the earliest manifestations.

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