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PHOTO QUIZ

Nodules on the tongue and thick lips

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CASE REPORT

A 40-year-old man presented to the Emergency Department with recurrent abdominal symptoms. He often presents with complaints of abdominal pain, constipation, and lack of appetite. In earlier visits, abdominal X-rays have sometimes shown a distended intestine and fluid levels. A computed tomography (CT) scan from an earlier admission showed mild diverticulitis. His medical history reveals medullary thyroid cancer at the age of 8 years, for which he underwent a thyroidectomy. Due to bilateral pheochromocytomas, the adrenal glands were removed at 34 and 35 years of age. He has been admitted several times for severe weight loss caused by the frequently recurring abdominal pain and has psychosocial problems resulting from this burden on life.

Figure 1. *Numerous yellow-white nodules on the tongue of a patient with a history of endocrine neoplasms*

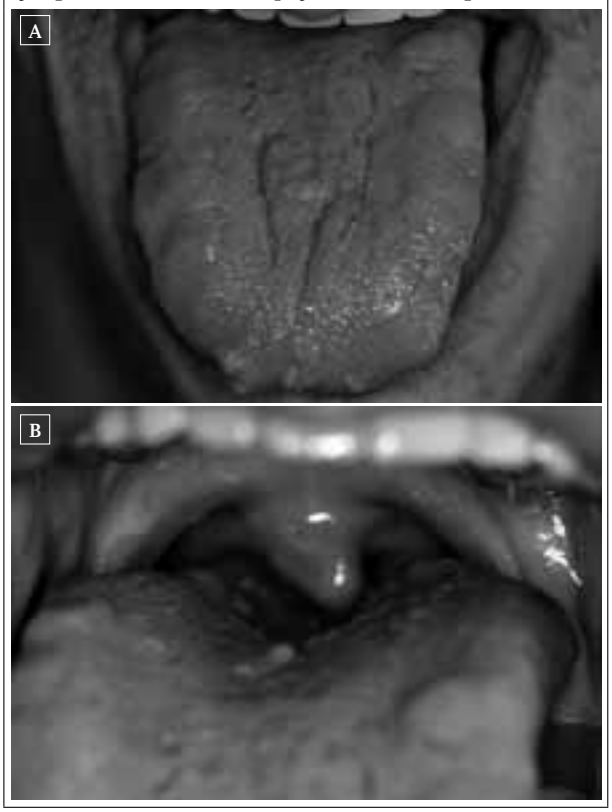
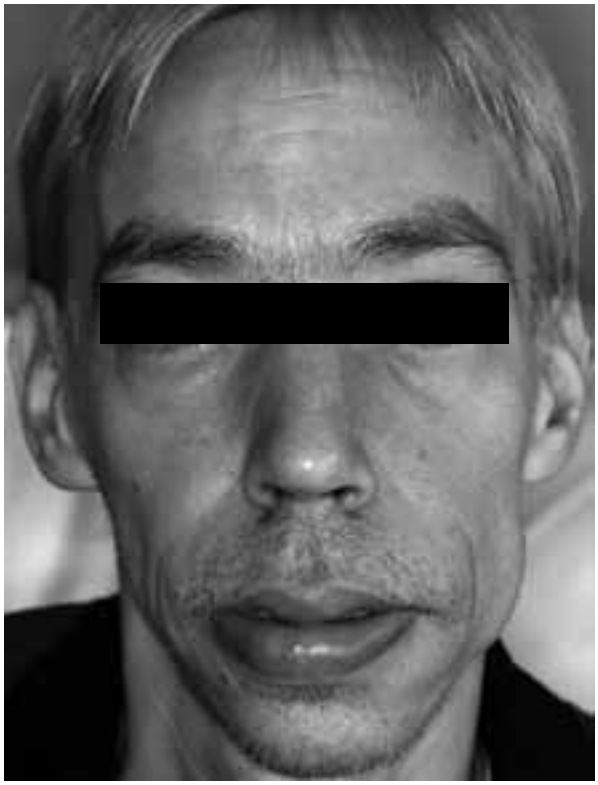


Figure 2. *A patient aged 40 years with Marfanoid facial appearance*



Apart from diffuse abdominal pain, physical examination shows nodules on the tongue, thick lips, and a Marfanoid appearance (*figures 1* and *2*). The nodules on the tongue and thick lips are pathognomonic for the underlying syndrome causing the gastrointestinal complaints.

WHAT IS YOUR DIAGNOSIS?

See page 234 for the answer to the photo quiz.

ANSWER TO PHOTO QUIZ (PAGE 231)

NODULES ON THE TONGUE AND THICK LIPS

DIAGNOSIS

This male patient, aged 40 years, has multiple endocrine neoplasia (MEN) syndrome type 2B.[1] MEN 2B is inherited as an autosomal dominant trait.[1] The mother and sister of this patient were also diagnosed with the syndrome. The characteristic features include medullary thyroid cancer (90% of patients), unilateral or bilateral pheochromocytomas (50% of patients), intestinal and mucosal ganglioneuromatosis (all patients), and a characteristic Marfanoid appearance. Our patient had medullary thyroid cancer and underwent a thyroidectomy at the age of 8 years. He also had bilateral pheochromocytomas, which were removed at the ages of 34 and 35 years.

Figure 1 shows the pathognomonic ganglioneuromatosis. Mucosal neuromas are the most consistent and distinctive feature of MEN 2B, appearing in all patients.[2] The presence of multiple mucosal neuromas is associated with diffuse intestinal ganglioneuromatosis, causing gastrointestinal problems (diverticulosis, persistent diarrhoea or constipation). The abdominal complaints can be a major burden on normal life. Currently, there are no treatment options for ganglioneuromatosis. This patient has been admitted repeatedly for severe weight loss and severe constipation due to gastrointestinal dysmotility, and was now admitted for severe constipation as well.

Figure 2 shows the characteristic Marfanoid facial appearance of this patient. On the left jaw line there is a lipoma present, which is not associated with the syndrome.

REFERENCES

1. Brandi ML, Gagel RF, Angeli A, et al. Guidelines for diagnosis and therapy of MEN type 1 and type 2. J Clin Endocrinol Metab. 2001;86:5658-71.
2. Carney JA, Go VL, Sizemore GW, Hayles AB. Alimentary-tract ganglioneuromatosis. A major component of the syndrome of multiple endocrine neoplasia, type 2b. N Engl J Med. 1976;295:1287-91.