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Medicine

MULTIPLE ENDOCRINE NEOPLASIA TYPE 2B SYNDROME: A RARE CASE PRESENTATION

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ABSTRACT Multiple endocrine neoplasia Type 2b is a rare syndrome caused by mutations in RET proto oncogene. It is a rare entity, which is found 1 in 30,000 people. We report a case of 35-year-old male patient who presented with 1 episode of syncope; had bumpy lips and mucosal neuromas over the tongue which on detailed biochemical and radiological investigations turned out to be MEN Type 2B syndrome.

KEYWORDS : multiple endocrine neoplasia, pheochromocytoma, medullary thyroid carcinoma, mucosal neuroma.

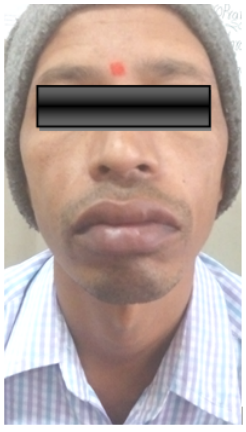
INTRODUCTION

MEN type 2 (MEN2) is an autosomal dominant syndrome that affects approximately one in 30,000 individuals1. Germline mutations in the proto-oncogene *RET* (*MIM 164761*), located on chromosome 10q11.2, are responsible for the three subtypes: MEN2A (80%; *MIM 171400*), MEN2B (5%; *MIM 162300*) and familial MTC[2]. MEN2B is characterized by MTC and pheochromocytoma plus marfanoid habitus, mucosal neuromas on the tongue, lips and subconjunctival areas and diffuse ganglioneuromas of the gastrointestinal tract . MEN type 2B (MEN2B) is the most distinctive and aggressive of the MEN2 variants[2].

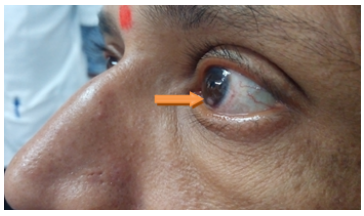
CASE STUDY

A 35 year old male patient, who had history of 1 episode of syncope leading to fall over abdomen 10 days back; was found to have an incidental supra-renal mass on abdominal ultrasonography (USG) for which he was referred to our hospital. He did not have any similar history of syncope in past. He did not complain of breathlessness on exertion, pedal edema, chest pain, palpitations. There were no similar complaints in any of the family members.

On examination, the patient had blood pressure of 150/100 mmHg and had enlarged bumpy lips (Pic. 1), mucosal neuromas over the conjunctiva (Pic. 2) and also on the tongue (Pic. 3). Though there was no obvious neck swelling, his neck ultrasonography showed multiple hypoechoic nodular lesions in bilateral thyroid lobes with macro and micro calcification and internal vascularity within, highly suspicious of neoplastic lesions which was further supported by raised serum calcitonin (1302 pg/ml; Ref <18.2). His contrast abdominal computed tomogram showed multiple retroperitoneal lymph nodes with left adrenal heterogenous enhancing lesion (Pic. 4) possibly an adrenal neoplasm which was suspected to be pheochromocytoma based on elevated free plasma metanephrine levels (204 pg/ml; Ref <65 pg/ml). Based on all the above features a diagnosis of MEN Type 2B Syndrome was made.

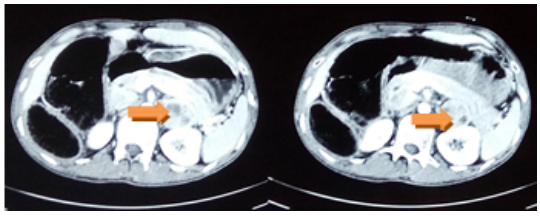


Pic. 1 (Enlarged bumpy lips).



Pic. 2 (Conjunctival neuroma).

Pic. 3 (Mucosal neuroma over tongue).



Pic. 4 (CECT abdomen showing heterogenous enhancing lesions in the left adrenal gland).

DISCUSSION

We report a case of multiple endocrine neoplasia (MEN Type 2B) who presented with mucosal neuromas, medullary carcinoma thyroid, pheochromocytoma. Williams and Pollock reported first case of MEN TypE 2B3. Chong first named this disease as MEN Type 2B. Patient with MEN Type 2B usually present in the first decade of life. Recent studies revealed age at diagnosis may range from 1 to 31 years, but our case was presented at the age of 35 years4. Among 100% of patients with MEN Type 2B develop mucosal neuromas in the lips, tongue and oral cavity, conjunctiva eye lids and within cornea5. Our case had mucosal neuromas over tongue and conjunctiva. Marfanoid habitus is present in 75% of MEN Type 2B as are skeletal abnormalities such as kyphosis, pectus excavatum and talipes supinatus. Our case did not have marfonid habitus. Medullary thyroid cancers are usually seen in 90-95% of MEN 2B and are commonly multiple, this malignant tumor appears in late teens or twenties. Hyperplasia of C-cells of thyroid and

hyperplasia of the adrenal medulla are thought to be pre malignant lesions of medullary thyroid cancer and pheochromocytoma. Our patient had medullary carcinoma thyroid with hypercalcitonemia. Medullary carcinoma of the thyroid in MEN Type 2B carries a poor prognosis compared with sporadic cases. Pheochromocytoma is usually manifested in 45-50% of patients with MEN Type 2B occurring during second and third decade of life. It is frequently multicentric and bilateral. Our patient had unilateral adrenal pheochromacytoma. 50% of MEN Type 2B are autosomal dominant, others are sporadic. Our case did not have any family history, and so it could be sporadic.

CONCLUSION

Finally, we report a rare case of MEN Type 2B who presented with pheochromocytoma, mucosal neuromas medullary carcinoma of the thyroid.

REFERENCES:

1. Diaz, R. E. & Wohllk, N. Multiple endocrine neoplasia: the Chilean experience. Clinics (Sao Paulo, Brazil) 67 Suppl 1, 7–11 (2012).
2. Brandi, M. L. et al. Guidelines for diagnosis and therapy of MEN type 1 and type 2. J Clin Endocrinol.Metab 86, 5658–5671 (2001).
3. Williams, E. D. & Pollock, D. J. Multiple mucosal neuromata with endocrine tumours: a syndrome allied to von Recklinghausen's disease. The Journal of pathology and bacteriology 91, 71–80 (1966).
4. Brauckhoff, M. et al. Multiple endocrine neoplasia 2B syndrome due to codon 918 mutation: Clinical manifestation and course in early and late onset disease. World Journal of Surgery 28, 1305–1311 (2004).
5. Pijnenburg-Kleizen, K. J., van Santen, H. M., Koolen, D. A. & Claahsen-van der Grinten, H. L. [Multiple endocrine neoplasia type 2B]. Ned. Tijdschr. Geneeskd. 159, A7719 (2015).