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Multiple endocrine neoplasia type 2B – the role of the ophthalmologist

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Background:

Multiple endocrine neoplasia type 2B (MEN2B) is rare (estimated prevalence between 1 in 600,000 to 1 in 4 million) but often fatal cancer syndrome. This hereditary autosomal-dominant condition is caused by activating germline mutations in the *RET* proto-oncogene. MEN2B syndrome can cause thyroid and parathyroid gland carcinoma, pheochromocytoma, and benign lesions such as multiple submucosal neuromas. Distinctive physical features are often seen in affected individuals, including marfanoid habitus, full lips and thickened eyelids. We aim to present patients with MEN2B diagnosed by ophthalmologists based on characteristic ocular findings.

Case presentation:

Between 2012 and 2022 at University Hospital Centre Zagreb MEN2B syndrome was suspected in two patients (aged 7 and 12 years) and confirmed by genetic testing. Molecular genetic analysis detected a mutation at codon 918 of the *RET* proto-oncogene present in 95% cases of MEN2B. Both patients were clinically examined, a detailed record of symptoms was made and a family history was taken, the necessary tests and procedures were performed. The patients had prominent corneal nerves and nodules located on eyelids, conjunctiva, lips and tongue so as characteristic facies. Serum calcitonin and intraocular pressure were elevated in both patients. Surgical treatment and histologic analysis confirmed medullary thyroid carcinoma. In one patient metastasis in the lymph nodes of the neck were found. There were no signs of pheochromocytoma.

Conclusion:

Characteristic findings of MEN2B include prominent corneal nerves in a clear stroma and multiple submucosal neuromas of the conjunctiva, eyelids, lips, and tongue. Ophthalmologists have a critical role to play in recognizing these signs, because the early diagnosis may be lifesaving. Higher awareness regarding the early non-endocrine signs of MEN2B could lead to earlier diagnosis, prevention of medullary thyroid cancer development and thus better prognosis.

Keywords:

medullary thyroid cancer, multiple endocrine neoplasia type 2B, ophthalmology, prominent corneal nerves, submucosal neuromas